Agenesis of the corpus callosum in preschool children

Wuebbens, Stephanie Ann, Ph.D.
The Ohio State University, 1991

Copyright ©1991 by Wuebbens, Stephanie Ann. All rights reserved.
AGENESIS OF THE CORPUS CALLOSUM
IN PRESCHOOL CHILDREN

DISSertation

Presented in Partial Fulfillment of the Requirements for
the Degree Doctor of Philosophy in the Graduate
School of the Ohio State University

By

Stephanie Ann Wuebbens, B.A., M.S

*****

The Ohio State University

1991

Dissertation Committee:
H. Leland
D. Hammer
G. Berntson

Approved by

Adviser
Department of Psychology
ACKNOWLEDGMENTS

My sincere appreciation to Dr. Henry Leland for his guidance, encouragement, and insight during the course of this research. Many thanks to Dr. David Hammer and Dr. Gary Berntson for their suggestions and guidance. Gratitude is expressed to Dr. Keith Yates and Dr. Eric Handler for their assistance and to Dr. Edward Kosnik for his medical expertise. Thank you to Dr. Donald T. Campbell for his thoughtful input on methodology and design.

To the parents of the children in this study, who were so willing to cooperate in the research and so eager for the information obtained, my thanks. To the agencies involved with the children's education, and to the teachers for their interest and assistance, my appreciation. A special thanks to my friends and colleagues who kept a sharp eye out for potential subjects.

To Greg, my love and continuing appreciation for the patience, understanding, and support through all the peaks and valleys.
VITA

August 14, 1959...... Born - Detroit, Michigan
1981.................. B.A., Capital University, Columbus, Ohio
1981-1984............. Case Manager, CompDrug VITA Youth Services, Columbus, Ohio
1984.................. M.S., The Ohio State University, Columbus, Ohio
1984-1985............. Psychology Intern, Midwest Neuropsychological Institute, Columbus, Ohio
1985-Present.......... Psychology Intern, Franklin County Board MR/DD, Early Childhood Education Center, Columbus, Ohio

FIELDS OF STUDY

Major Field: Psychology


Studies in Developmental Psychology. Professors Dorothy Jackson, Dale Blythe, Felicisima Serafica, Gerald Winer, John Gibbs.


Studies in Family Relations and Human Development. Professors Barbara Newman, Joseph Mullan.
# TABLE OF CONTENTS

ACKNOWLEDGEMENTS

VITA

LIST OF TABLES

LIST OF FIGURES

CHAPTER PAGE

I. INTRODUCTION ................................. 1
   Statement of the problem ......................... 10
   Objective .......................................... 11
   Research questions ................................ 11
   Definition of terms ................................ 12

II. REVIEW OF THE LITERATURE ..................... 13
   Development of the cerebral commissures .......... 13
   Callosal agenesis .................................. 16
   Associated anomalies ................................ 17
   Probst's bundles .................................... 17
   Other structural anomalies ....................... 18
   Associated neurological anomalies ............... 21
   Shapiro syndrome ................................... 22
   Andermann syndrome ................................ 23
   Holoprosencephaly .................................. 23
   Prevalence ......................................... 25
   Etiology .......................................... 26
   Clinical features ................................... 29
   Cognitive development ............................. 30
   Language development .............................. 32
   Visual functioning ................................. 37
   Visual-motor integration and bimanual
      coordination ..................................... 40
   Bimanual transfer of learning .................... 44
   Kinesthetic functioning ........................... 45
   Summary ............................................ 46

III. METHODOLOGY .................................. 48
   Subjects and setting .............................. 48
VI. DISCUSSION .......................................... 156

General Findings........................................ 156
Patterns of Developmental Delay ..................... 157
Specific Developmental Delays ........................ 160
  Visual-motor functioning........................... 161
  Language skills.................................... 162
Age and Developmental Delay ........................... 163
Implications of the Study ............................. 165
Implications for Education of Young Acallosal Children ............................. 167
Limitations of the Study............................. 168
Future Research....................................... 170
Conclusions........................................... 171

LIST OF REFERENCES.................................... 173
LIST OF TABLES

<table>
<thead>
<tr>
<th>TABLE</th>
<th>PAGE</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Subject characteristics</td>
<td>49</td>
</tr>
<tr>
<td>2. Age at diagnosis and symptoms</td>
<td>58</td>
</tr>
<tr>
<td>3. Complications at birth</td>
<td>60</td>
</tr>
<tr>
<td>4. Seizure type and age at onset</td>
<td>61</td>
</tr>
<tr>
<td>5. Congenital anomalies as noted in medical records</td>
<td>63</td>
</tr>
<tr>
<td>6. McCarthy results</td>
<td>65</td>
</tr>
<tr>
<td>7. Bayley results</td>
<td>66</td>
</tr>
<tr>
<td>8. Adaptive behavior results</td>
<td>68</td>
</tr>
<tr>
<td>9. Verbal measures</td>
<td>72</td>
</tr>
<tr>
<td>10. Visual-motor tasks</td>
<td>73</td>
</tr>
<tr>
<td>11. Case 2: Previous developmental levels</td>
<td>85</td>
</tr>
<tr>
<td>12. Case 3: Previous developmental levels</td>
<td>87</td>
</tr>
<tr>
<td>13. Comparison of functioning levels: subjects with agenesis and partial agenesis matched by age</td>
<td>89</td>
</tr>
<tr>
<td>14. Case 1: Summary of results</td>
<td>102</td>
</tr>
<tr>
<td>15. Case 2: Summary of results</td>
<td>113</td>
</tr>
<tr>
<td>16. Case 3: Summary of results</td>
<td>120</td>
</tr>
<tr>
<td>17. Case 4: Summary of results</td>
<td>126</td>
</tr>
<tr>
<td>18. Case 5: Summary of results</td>
<td>134</td>
</tr>
</tbody>
</table>
19. Case 6: Summary of results ......................... 139
20. Case P7: Summary of results ......................... 147
21. Case P8: Summary of results ......................... 154
# LIST OF FIGURES

<table>
<thead>
<tr>
<th>FIGURE</th>
<th>PAGE</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. McCarthy results</td>
<td>67</td>
</tr>
<tr>
<td>2. ABSI results: Moderate to severe delays</td>
<td>70</td>
</tr>
<tr>
<td>3. ABSI results: No significant to mild deficits</td>
<td>71</td>
</tr>
<tr>
<td>4. Patterns of completion: Wallin pegboard</td>
<td>75</td>
</tr>
<tr>
<td>5. Scatterplot: Comparison of age and measured intelligence levels</td>
<td>77</td>
</tr>
<tr>
<td>6. Scatterplot: Comparison of measured intelligence and number of complications</td>
<td>78</td>
</tr>
<tr>
<td>7. Scatterplot: Comparison of adaptive behavior levels and number of complications</td>
<td>79</td>
</tr>
<tr>
<td>8. Change in measured intelligence level over time: Case 1</td>
<td>81</td>
</tr>
<tr>
<td>9. Case 1: Comparison of developmental levels across domains</td>
<td>82</td>
</tr>
<tr>
<td>10. Case 2: Comparison of developmental levels across domains</td>
<td>84</td>
</tr>
<tr>
<td>11. Case 3: Comparison of developmental levels across domains</td>
<td>86</td>
</tr>
<tr>
<td>12. ABSI results: Comparison of subjects with partial agenesis</td>
<td>89</td>
</tr>
</tbody>
</table>
CHAPTER I
INTRODUCTION

While agenesis of the corpus callosum has been known for a century, having first been reported in the late 1800's (Field, Ashton & White, 1978), the scientific study of the corpus callosum is a recent development in history. Clarke and O'Malley's (1968) survey of original writings on the structure and function of the brain indicate that the corpus callosum has long been the subject of inquiry. They found that Galen mentioned the corpus callosum back in the 2nd Century A.D. In the 16th century, Piccolomini was one of the first to make clear distinctions between the grey and white matter. His conception of the function of the corpus callosum is that of a structural border between the grey matter and the white matter. Vesalius suggested a structural function in 1543. He saw the corpus callosum as a connection between the hemispheres, supporting the septum and the fornix so that these structures would not crush the ventricles (Clarke & O'Malley, 1968).
Probst (1973) and Tomasch (1954) note that the popular view during 17th to 19th centuries was that the corpus callosum was the source of the soul. Thomas Willis, toward the end of the 17th Century, mentioned the corpus callosum when discussing memory:

Regarding the former [common sense] we notice...a sensory impression, like an optical appearance or like the undulation of water, is carried more inwardly, turning towards the corpora striata...if this impression...crosses through the corpus callosum, it is succeeded by imagination; then, if that same flowing of the spirits strikes against the cortex of the brain, as its farthest shore, it impresses on it the image or character of the sensible object; when thereafter the same image is reflected, it arouses the memory of the same thing" (Willis, 1672, p.72-73, excerpted by Clarke & O'Malley, 1968, p. 334).

Clarke and O'Malley (1968) note that when Willis, and the other anatomists of that time, refer to the corpus callosum, they refer to "all parts of the white matter, in addition to the structure known by this name today" (p. 472). It was not until the invention of the microscope in
the 19th century that progress was made in understanding specific areas of the white matter (Clarke & O'Malley, 1968).

Felix Vicq d'Azyr in 1784, was one of the first to note, "It seems to me that the commissures are intended to establish sympathetic communications between the different parts of the brain" (pp. 535-536, excerpted in Clarke & O'Malley, 1968, p. 592)

Probst (1973) notes that as recently as 1907, Spitska debated about the relationship between intelligence and the number of callosal fibers. The 1920's and 1930's are seen by Probst as a time of accumulation of data, and the first indications of disturbances of interhemispheric transfer.

Before the mid-1950's, virtually nothing was known about the function of the corpus callosum in spite of spirited debate (Chiarello, 1980). McCulloch (1940) stated that the only known function of the corpus callosum was to transmit seizures from hemisphere to hemisphere.

Akelaitis (1944) asserted that provided no other damage existed, transsection of the corpus callosum and the anterior commissures produced no functional symptoms. His studies influenced the direction of many researchers in the 1940's and 1950's (Sperry, 1968). Further, Lashley is reported to have sarcastically noted during this period
that the principle function of the corpus callosum is to keep the hemispheres from sagging (Sperry, 1962).

In contrast, textbooks of the time attributed orientation, speech, visual and auditory functions to the corpus callosum (Tomasch, 1954). Tomasch (1954) noted a divergence of theories on the origins and functions of the corpus callosum. Hebb (1949) theorized that sensory events are integrated bilaterally, with each half of the brain mediating perception and facilitating responses, with the corpus callosum and other commissures coordinating that integration. He suggested further that bilateral development is dependent on the corpus callosum.

Much of what is known about the corpus callosum is derived from commissurotomy ("split-brain") studies, first with animals, and later with humans for the treatment of intractable seizure activity. Chiarello (1980) submits that current theories emphasize the importance of the corpus callosum in interhemispheric integration as the major link between functionally discrete hemispheres.

Harrington (1987) noted that there is early evidence of what is known as the split-brain syndrome. He reports that in the 1880's, Miles had a patient with a tumor of the corpus callosum who had difficulties with language processing and took an unusually long time to answer
questions. Further, he noted that Liepmann did a detailed case study in 1900 of a man with lesions of the left parietal region and the corpus callosum which resembled the split brain syndrome of persons with transsected corpus callosums, providing the first evidence that it functioned as a pathway for interhemispheric information.

When Myers and Sperry began to reexamine cats with callosum sections and later when more dramatic results with monkeys were revealed, the communicative nature of the corpus callosum began to be taken seriously (Gazzaniga, Bogen & Sperry, 1962; Sperry, 1962). Still, with the initial studies of transsected patients, the relative lack of effect on everyday behavior raised questions as to whether the split-brain syndrome would be observed in humans (Sperry, 1968). The effects of callosal transsection in humans are now well documented.

A wide variety of functions are currently ascribed to the corpus callosum, including the transfer of cognitive, perceptual, and emotional information between the hemispheres (Bogen & Bogen, 1988; Cronin-Golomb, 1986), with recent interest in the types of information that transfer subcortically (Cronin-Golomb, 1986). A current focus of corpus callosum research is its function in schizophrenia, particularly in epileptic psychosis (Conlon

Gazzaniga (1970) notes that studies of callosal agenesis have both advanced and confused discussion of function. He finds that both commissurotomy research and research with acallosals offers few insights. Rather, it increases the problems and raises more questions.

Agenesis of the corpus callosum is a relatively rare neurological condition (Andermann, 1981; Ettlinger, 1977; Myrianthopolous, 1987). The effects of agenesis may be mild, or may be associated with syndromes or other conditions that interfere with a child's functioning.

Early evidence of persons with agenesis studied at autopsy by Ireland (1885) contributed to the confusion about the function of the corpus callosum. He retrospectively found three acallosal subjects to have been free of affective disturbance, splitting of the personality, or intellectual deficits, and concluded that the brain could function normally without a corpus callosum.

Frequently, the symptoms in acallosals have been subclinical, and many of the earliest cases studied were confirmed only after autopsy (Field et al., 1978). In the
literature, at least two cases (Bruyer, Dupuis, Ophaven, Rectem & Reynaert, 1985; Buchanan, Waterhouse, & West, 1980) have had so few symptoms that the discovery of agenesis was made by chance.

Many of the acallosals who have been studied have tended to function in the mild range of mental retardation to the average range of intellectual functioning. There are no specific patterns of deficits established, due to the small number of acallosals studied, the need for reliable measures of associated features, and the fact that many observed deficits are likely to be the result of associated developmental anomalies rather than as a result of the agenesis (Ettlinger, 1977; Field et al., 1978; Loeser & Alvord, 1968).

In spite of recent interest, few persons with congenital agenesis are studied. Jeeves and Temple (1987) propose two explanations. First, many cases may go undetected, although with increased use of computerized diagnostic tools, more cases are diagnosed and studied. Second, a proportion of cases found are mentally retarded, and fewer of these cases are researched.

While studies of adult and adolescent acallosals are infrequent, investigations of children, particularly those of preschool age, are more uncommon. Those who have
studied or reviewed studies of children suggest that younger acallosals exhibit more symptoms than adults, including more clumsiness, speech delays and cognitive delays (Chiarello, 1980; Teeter & Hynd, 1981). Further, there is evidence that as development progresses, acallosals compensate for deficits. It is hypothesized that compensatory patterns of organization occur in the absence of the corpus callosum (Field et al., 1978). Sauerwein, Lassonde, Cardu and Geoffroy (1981) and Teeter & Hynd (1981) hypothesize that as the child matures, early deficits become less apparent.

Animal research has generally been limited to study of transsection of the corpus callosum (Sperry, 1961; Gazzaniga, 1989) although agenesis of the corpus callosum is a known anomaly in the BALB mouse strain (Geschwind & Galaburda, 1987), and some studies utilize chemical hemispheric depression (Bogen & Bogen, 1988). Early animal research suggested that transfer of learning depended on an intact corpus callosum, and that normal cognitive processes continued independently in each hemisphere (Sechzer, Folstein, Geiger, & Mervis, 1977). More recently, animal research indicates that animals tend to have significant structural overlap (Demeter, Rosene, & Van Hoesen, 1990).
particularly in terms of visual or spatial functions, that humans may not (Foxman, Oppenheim, Petito, & Gazzaniga, 1986; Gazzaniga, 1970, 1989; Martin, 1985).

Studies of callosal transsection, and studies of acallosal adults do not supply us with data regarding the age at which the corpus callosum begins to function in interhemispheric transfer, nor do they tell us about the role of the corpus callosum in normal brain development (Sechzer et al., 1977).

Study of callosal agenesis is important, in terms of the information it provides regarding interhemispheric transmission of information. Chiarello (1980) suggests that from a careful comparison of acallosals and persons with comissurotomies, we can determine which functions cannot be compensated.

Further, study of the corpus callosum provides information regarding the mechanisms of adaptation to cerebral anomalies. Both the transsection patients and those with congenital agenesis appear to have a mode of compensation for their respective deficits. Transsection patients appear to compensate through cross-cueing. Cross-cueing and structural plasticity, including alternate pathways (i.e. the anterior commissure) and bilateral development of language, are proposed as the mechanisms of
the ability of the acallosals to compensate (Ettlinger, 1977; Gazzaniga, 1970; Gott & Saul, 1978).

As recently as 1984, it was noted that:

"The question arose—and still remains— if the corpus callosum is of such vital importance for the normal interhemispheric transfer and integration of information, how is it that those born without the major neocortical commissural pathway may appear to lead normal lives without consequence of its absence?"

(Jeeves, 1984, p. 235).

**Statement of the problem**

Agenesis of the corpus callosum is a low incidence structural deficit. We are just beginning to understand the importance of this brain structure, and a study of individuals with this defect can provide more information on its function. Further, study of young children with agenesis of the corpus callosum can yield data to aid in the understanding of ways in which the function of the corpus callosum changes over time, and how children who are acallosal compensate for their deficit. There are suggestions in the literature that younger children show evidence of deficits for which they later compensate, yet there are only a handful of studies of acallosal children.
Objective

The purpose of this study is to assess whether preschool children who have been identified as having agenesis of the corpus callosum exhibit a pattern of delays that relate to the functioning of the corpus callosum. Further, I wish to provide additional information about the functioning of preschool children with agenesis of the corpus callosum, to better understand the role of the corpus callosum during development and the nature of their compensation for deficits.

Research questions

1. Will children with absence of a corpus callosum have an identifiable, specific pattern of developmental delays.

2a. Will acallosal children display delays in visual-motor tasks and language.

2b. Will acallosal children show greater delays in visually aided bimanual tasks than bimanual tasks completed without visual input.

3. Will younger children be more likely to exhibit identifiable delays than older children with agenesis.

4. Are there differences in patterns of delays that could be a result of individual differences in addition to extent of associated anomalies.
Definition of terms

Acallosal: A person with congenital absence of the corpus callosum.

Bimanual: Use of both hands.

Callosal Agenesis: Defect or absence of the callosal structures (Friel, 1974).

Corpus Callosum: An arched mass of white matter within the longitudinal fissure which is composed of transverse fibers connecting the cerebral hemispheres.

Holoproencephaly: Failure of the medial cleavage of the proencephalon with a resulting defect in midline facial development. Sometimes associated with trisomy 13-15 or trisomy D-1. Characteristics include low-set ears, bilateral cleft lip and palate, microcephaly, ocular abnormalities, seizures and mental retardation (Friel, 1974). Probst (1973) emphasizes the need to differentiate this condition from agenesis of the corpus callosum.
CHAPTER II
REVIEW OF THE LITERATURE

Development of the cerebral commissures

During normal embryological development, the precursor of the brain is the neural tube. By approximately the third week, the neural plate has formed on the dorsal side of the embryo. The neural plate begins to fold, forming the neural groove, which develops into the neural tube through the closure of the neural folds. The cells of the neural tube multiply quickly, and eventually develop into the structures of the brain (Heimer, 1983).

Commissural fibers begin to form in the embryonic rhombencephalon during the fourth week (O'Rahilly & Muller, 1987). The next commissures to form include the anterior, hippocampal, posterior and habenular commissures (Heimer, 1983; O'Rahilly & Muller, 1987). Heimer (1983) describes the posterior commissure as a complex bundle of fibers that join proximal areas. These include the pretectal area and portions of the ocular motor complex. The posterior commissure is nearly always present in cases of callosal agenesis, due to independent development sites (Loeser & Alvord, 1968; Probst, 1973).
The habenular commissure develops after the posterior commissure (O'Rahilly & Muller, 1987), and sits above the pineal recess. It is developed from the diencephalon which connects the habenular complex and other subcortical regions with the corresponding contralateral areas (Heimer, 1983).

The anterior lamina reuniens is the site of the anterior commissure, which develops by 11-12 weeks, and connects the olfactory nuclei (Heimer, 1983), the amygdaloid nuclei and areas of the temporal cortex (Probst, 1973).

The posterior section of the lamina reuniens is the site of the hippocampal commissure, which appears at approximately 11 weeks (Probst, 1973). The hippocampal commissure consists of commissural fibers which connect the hippocampal formations between the crua fornices and is located beneath the splenium, "to which it is intimately attached" (Heimer, 1983, p. 29).

By the 39th day of development, the commissural plate forms in the upper part of the terminal plate (Probst, 1973). It is the site of development of the anterior and hippocampal commissures and the corpus callosum (Loeser &
Alvord, 1968). The commissural plate extends from the precursor of the optic chiasm to the boundary between the diencephalon and the telencephalon, and serves as a passive tract that facilitates the crossing of commissural axons (Probst, 1973). Before the fibers cross, at approximately 60 days, the commissural plate thickens, a medial indentation develops, and the gap is filled in by commissural plate cells. The commissural fibers begin to cross the commissural bed at 12-13 weeks (Aicardi, Chevrie & Baraton, 1987; Probst, 1973; Rakic & Yakovlev, 1968). Once the commissuration across the plate begins, it continues throughout the majority of the fetal period (O'Rahilly & Muller, 1987).

The corpus callosum has not been observed in embryos smaller than 55 mm (Rakic & Yakovlev, 1968). Once commissuration begins, the genu and body are formed by rostral and caudal growth during weeks 14 through 16. The timing of the development of the splenium and rostrum is debated (Aicardi et al., 1987; Probst, 1973).

By 17-20 weeks the corpus callosum generally resembles the shape of the adult (Chiarello, 1980). The corpus callosum continues to grow through the prenatal period (Atlas, Zimmerman, Bilaniuk, Rorke, Hackney,
Goldberg, & Grossman 1986; Chiarello, 1980), with growth continuing postnatally up to two years of age (Aicardi et al., 1987) and beyond (Rakic & Yakovlev, 1968).

Myelination of the corpus callosum begins after other fiber tracts. Yakovlev and Lecours (1967) find that myelination begins postnatally between the second week and the second month of life. Myelination is generally complete by the age of 10, although not all fibers are myelinated (Tomasch, 1954).

**Callosal agenesis**

Structurally, Probst sees agenesis of the corpus callosum as comprising one group, however in terms of etiology, the group is heterogeneous. Some cases result in agenesis with no associated anomalies due to an isolated lesion associated with the commissural plate. Other cases include agenesis as a part of a syndrome of malformations, and are more likely to be associated with a generalized lesion of early brain structures (Probst, 1973).

The degree to which the corpus callosum is developed depends on the timing of fetal insult in the developmental sequence and the nature of the damage (Atlas et al., 1986; Chiarello, 1980; Probst, 1973). Fetal insult at an early stage is likely to result in agenesis of the anterior commissure, the hippocampal commissure, and the corpus callosum, with no commissural axons extending from the cortex (Chiarello, 1980; O'Rahilly & Muller, 1987).
If the insult occurs after 11 weeks, the anterior commissure and developing hippocampal commissure are spared, resulting only in callosal agenesis (Probst, 1973). Agenesis without other complications is relatively rare; some involvement of the anterior commissure is frequently observed (Atlas et al., 1986, Chiarello, 1980). Damage that occurs after the eleventh week represents failure of the commissuration process or dysgenesis (Rakic & Yakovlev, 1968). The fibers grow from the cortex, reach the median plate, and instead of crossing, grow in both anterior and posterior directions, forming Probst's bundles (O'Rahilly & Muller, 1987).

Associated anomalies

Probst's bundles

The presence of Probst's bundles, found on the medial wall of each ipsilateral hemisphere, is thought to be a reflection of the brain's ability to compensate. They connect ipsilateral rather than contralateral structures (Chiarello, 1980; Probst, 1973). Chiarello (1980) notes that similar longitudinal bundles are found in the strain of acallosal mice.
Probst's bundles are formed when callosal fibers grow from the cortex, reach the median plate, and instead of crossing, grow in anterior and posterior directions (O'Rahilly & Muller, 1987). Kendall (1983) notes that these fibers are present in all but the most complicated cases. The fibers run sagittally from the frontal (Atlas et al., 1986) to the occipital lobe (Atlas et al., 1986; Probst, 1973), where the fibers split off into the tapetum with lateral outgrowths to the forceps (Probst, 1973). With partial agenesis, Probst's bundles are found only in areas where corpus callosum does not exist (Chiarello, 1980). In these cases, the Probst's bundles tend to also contain short fibers which connect them to the callosal rudiment (Probst, 1973). O'Rahilly and Muller (1987) suggest that persons who also lack an anterior commissure would not develop Probst's bundles due to the timing of the insult.

**Other structural anomalies**

Atlas and colleagues (1986) note that the limbic system is likely to be affected, due to the proximity to the corpus callosum. In their sample, all ten of the subjects studied with Magnetic Resonance Imaging (MRI) had limbic system anomalies.
One frequently associated anomaly is distortion in the shape of the lateral ventricles (Chiarello, 1980; Lewis, Reveley, David & Ron, 1988; Probst, 1973). This distortion was one of the important diagnostic features of agenesis, before the availability of MRI. Due to the expense of the latter procedure, ventricle shape remains an important diagnostic criteria. Prenatal diagnosis using ultrasound continues to rely on ventricle shape (Filly, 1990).

The type of distortion of the lateral ventricles vary, but similarities exist. Dilation of the posterior horns is common (Chiarello, 1980). There may be an upward extension of the lateral ventricles into the frontal and parietal areas (Lewis et al., 1988). The ventricles tend to be separated, particularly in the anterior area where the cingulate gyrus may infiltrate (Aicardi et al., 1987; Loeser & Alvord, 1968; Probst, 1973). The anterior horns and bodies tend to be constricted into narrow slits or clefts, which resemble bulls horns (Probst, 1973).

The third ventricle may have cystic dilations (Lewis et al., 1988; Loeser & Alvord, 1968; Probst, 1973), and in cases of partial agenesis the septum pellucidum often contains cysts (Lewis et al., 1988). A wide interhemispheric fissure, open to the third ventricle is a common finding (Atlas et al., 1986).
Complete agenesis of the corpus callosum often includes agenesis of the hippocampal commissure (Lewis et al., 1988). The anterior commissure is also sometimes absent in agenesis (Aicardi et al., 1987; Loeser & Alvord, 1968; Probst, 1973). When present, it may be normal or hypertrophied (Atlas et al., 1986; Loeser & Alvord, 1968). There is evidence to suggest that hypertrophy is the result of callosal fibers crossing at this site (Loeser and Alvord, 1968).

A radial pattern of sulci on the medial area of the hemispheres and abnormally shaped posterior ventricles have also been observed in some cases (Chiarello, 1980). Loeser & Alvord (1968) comment that this pattern is always present in agenesis, and is not, as some have suggested, a fetal pattern. In partial agenesis, this radiating pattern is present in areas where the corpus callosum is absent. Their hypothesis is that this pattern may be due to pressure from cortical folding during development, and the pattern reflects the direction of growth.

Agyria is sometimes associated with agenesis of the corpus callosum (Aicardi et al., 1987; Probst, 1973). Midline anomalies, such as cleft palate, are often found (Lewis et al., 1988). Atlas and his colleagues (1986) noted the presence of interhemispheric cysts in 2 of 7 patients
they studied. Microgyria, subependymal cysts, and heterotopias are also reported (Loeser & Alvord, 1968).

**Associated neurological anomalies**

People who are identified as agenic tend to present symptoms that indicate the need for neurological examination with tools such as MRI (Jeeves, 1984). Hydrocephalus and seizure activity are two neurological anomalies most commonly associated with agenesis of the corpus callosum. Six of 29 cases reviewed by Chiarello (1980) were diagnosed as hydrocephalic. One of the two subjects reported by Field, Ashton, and White (1978) had hydrocephalus. An infant reported by Teeter & Hynd (1981) had suspected but unconfirmed hydrocephalus. A review of autopsied cases found that 44% of agenesis cases had evidence of hydrocephalus (Jellinger, Gross, Kaltenback & Grisold, 1981).

Of the 29 cases reviewed by Chiarello (1980), 14 had some type of seizure activity, and three had borderline abnormalities on the EEG. The subject discussed by Milner (1982) developed grand mal seizures at the age of 19. One of the two subjects reported by Sauerwein, Lassonde, Cardu, and Geoffroy (1981) had a dysrhythmic EEG but no reported seizure activity. Loeser & Alvord (1968) note that agenics tend to have nonspecific EEG irregularities.
Other commonly reported anomalies include macrocephaly (Bruyer, Dupuis, Ophaven, Rectem, & Reynaert, 1985; Field et al., 1978) and microcephaly (Chiarello, 1980; Jellinger, et al., 1981). Probst (1973) states that these conditions are a subset of dyscranias which he considers to be common in agenesis. He discusses dyscrania in agenesis of the corpus callosum as a manifestation of cerebral dysplasia. It emerges as abnormal size (macro- or microcephaly), asymmetric distortions, fontanelle and suture anomalies, hypertelorism, facial dysplasias, and midline skull defects.

**Shapiro syndrome**

Aicardi et al. (1987) describe Shapiro syndrome as a rare condition characterized by episodic hypothermia and diaphoresis in persons with agenesis of the corpus callosum. Generally, onset is in middle to late adulthood, although two cases have been reported before the age of two. During the episodes body temperature drops and is associated with fatigue, lethargy, confusion and mutism. Ataxia and tremors are also features. Death has occurred during these episodes in some cases. The source may be hypothalamic lesions rather than agenesis (Aicardi et al., 1987). Other hypotheses include seizures and changes in metabolic set point (Sanfield, Linares, Calahan, Forrester,
Halter, & Rosen, 1989). Johnson and Jones (1985) describe a case with a single episode of hypothermia associated with status epilepticus and significant metabolic changes. Recently, there is evidence that treatment of increased Norepinephrine levels may control this condition (Sanfield et al., 1989).

Andermann syndrome

Andermann syndrome as described by Aicardi et al. (1987) is characterized by callosal agenesis, mental retardation, distinctive facial features, and peripheral neuropathy. There appears to be a strong consanguinity component, as all known cases come from the same region in Quebec and can be traced to a common ancestor. Transmission is apparently autosomal recessive. Neuropsychological evaluation is described as difficult due to mental retardation and sensory loss. Motorically, the children deteriorate and are wheelchair bound by early adolescence.

Holoprosencephaly

Holoprosencephaly is due to failure of the midline cleavage of the forebrain during embryonic development (Jellinger et al., 1981). Various writers debate whether to include holoprosencephaly in discussions of callosal agenesis.
Probst (1973) notes that agenesis of the corpus callosum has been considered a continuous spectrum that depends on timing of the insult. He acknowledges that the developmental timing is significant to the nature of the deficit, but states there is not a constant relationship between the effect of the teratogen and the type of malformation that occurs. He proposes a classification system based on the degree of absence of the corpus callosum and includes holoprosencephaly and hypogenesis (abnormally thin corpus callosum) in his system. He distinguishes holoprosencephaly from agenesis of the corpus callosum, but considers the two conditions similar.

Loeser and Alvord (1968) also consider the two part of the same continuum. They note that some of the anomalies attributed to agenesis by early researchers may be because they were studying holoprosencephaly. Geschwind and Galaburda (1987) define agenesis as a failure of the two sides of the neural tube to fuse normally, and holoprosencephaly as the opposite extreme, where the forebrain is fused together, with a single ventricle. Aicardi et al. (1987) note that holoprosencephaly is one of the syndromes that includes agenesis of the corpus callosum but note that it has a different significance when the hemispheres have separated in the normal manner. Fitz
(1983) notes that a common misdiagnosis has been to mistake semilobar holoprosencephaly with callosal agenesis accompanied by midline cyst, when the two conditions have significant structural differences. Careful observation (Fitz, 1983) and more accurate diagnostic tools (Aicardi et al., 1987) are necessary for differential diagnosis.

Prevalence

Myrianthopolous (1987) considers agenesis of the corpus callosum a rare occurrence. He notes that reliable figures regarding incidence and prevalence are unavailable because it is a "malformation compatible with life" (p. 61). He cites a World Health Organization study which reported 1 case (with multiple malformations) in 20,074 live births in Czechoslovakia and 1 stillborn case in 24,700 births in Mexico City.

Other recent studies of prevalence estimate that agenesis occurs in 0.5-0.7% of the general population (Andermann, 1981; Ettlinger, 1977; Jellinger et al., 1981). Studies of autopsied cases suggest prevalence levels of 2.2% to 26% in mentally retarded populations (Andermann, 1981; Jellinger et al., 1981). Jeret, Serur, Wisniewski and Fisch (1985-86) suggest that due to factors in the institutions in the 1970's, there may have been an overestimation of agenesis in the mentally retarded
population. Their estimation of prevalence is 2.3% in the developmentally disabled population.

**Etiology**

The causal factors involved in agenesis are seen as multifactorial (Andermann, 1981). At present, no specific etiology has been established for those cases of agenesis of the corpus callosum which are not related to a syndrome (Aicardi et al., 1987, p. 152).

Environmentally related effects are suggested as potential factors (Aicardi et al., 1987; Andermann, 1981; Ettlinger, 1977). Specific maternal factors noted have included rubella, congenital toxoplasmosis, alcohol effects, and one mother with severe diabetes (Andermann, 1981).

Biochemical factors have been suggested, such as abnormalities in amino acids, and lipid deficits in white matter. One hypothesis for the latter finding is a localized metabolic disturbance (Menkes, Phillipart, & Clark, 1964). Another proposed biochemical factor is an early disruption in the development of the massa commisuralis may produce chemical changes that interferes with corpus callosum development (Chiarello, 1980). Barkovich and Norman (1988) concur that the massa commisuralis is closely involved in the ability of the fibers to cross.
Vascular lesions, which may cut off blood supply, have the potential to prevent structural development of the corpus callosum (Chiarello, 1980; Ettlinger, 1977). Trauma is also a potential cause (Aicardi et al., 1987; Ettlinger, 1977).

The presence of a midline cyst may also interfere (Chiarello, 1980; Loeser & Alvord, 1968). Others consider cysts a result of the etiological factors that produced the agenesis (Probst, 1973).

Genetic factors in agenesis have been studied by several researchers. Agenesis of the corpus callosum has occasionally been observed to run in families (Andermann, 1981; Ettlinger, 1977). Lewis and his colleagues (1988) note that the subjects studied by Andermann (1981) had an autosomal recessive link.

Lynn and his colleagues (Lynn, Buchanan, Fenichel & Freemson, 1980) studied a father and son with autosomal dominant traits. Lassonde and her colleagues have extensively studied a brother and sister (Lassonde, Sauerwein, McCabe, Laurencelle, & Geoffroy, 1988). A third sibling has recently been identified (Lassonde, Bryden, & Demers, 1990).
The brothers described by Lachiewicz and colleagues (Lachiewicz, Kogan, Levitt, & Weiner, 1985) are thought to have a single gene anomaly, perhaps autosomal recessive or X-linked recessive. One brother had total agenesis with hydrocephaly, the other partial agenesis with macrocephaly, and both boys had associated features that included ureteroceles and facial dysplasias.

Menkes and his colleagues (1964) describe a X-linked recessive form of agenesis characterized by the development of seizures in the first few days of life, severe developmental delay and frequently, death prior to two years of age. Associated anomalies include polymicrogyria and white matter heterotopias, with Probst's bundles usually absent.

Agenesis of the corpus callosum may be present in a variety of chromosomal anomalies (Atlas et al., 1986; Chiarello, 1980). These include trisomy 18, trisomy 13 (Andermann, 1981; Ettlinger, 1977), trisomy 8 (Aicardi et al., 1987; Andermann, 1981), Klinefelter syndrome, translocation of chromosome 2 and the B group, and asymmetric 4-15 translocation (Andermann, 1981). Aicardi et al. (1987) note that some of the chromosomal syndromes occasionally include agenesis, while others, such as Trisomy 8 mosaic regularly include partial or total
A strain of acallosal mice with a clear genetic connection exists (Chiarello, 1980; Ettlinger, 1977). Barkovich and Norman (1988) state that the nature of the insult is less important than the timing of that insult and the genetic vulnerability of the fetus.

Clinical features

It has been said that nature's experiments are badly designed (Hebb, 1949). This is certainly the case when studying agenesis of the corpus callosum. The presence of damage to other areas of the brain is one factor that may influence the results of studies (Atlas et al., 1986; Field et al., 1978). Concurrent neurological involvement is frequently associated with acallosals. Atlas and his colleagues (1986) list 33 structural abnormalities that were associated with their subjects.

A second confounding factor is that differences have been found between subjects with total and partial agenesis. Field and her colleagues (1978) found that the subject with total agenesis had more selective deficits than the subject with partial agenesis. The issue that has not been specifically addressed in the literature, is that studies done prior to the development of MRI may have been testing partial agenesis subjects rather than total agenesis. The MRI technology has improved the ability to
study brain structures (Atlas et al., 1986) and allows for detection of minor callosal abnormalities (Davidson, Abraham & Steiner, 1985).

Finally, there is the issue of the age at which acallosal subjects are studied. It is hypothesized by some that compensation and plasticity are two factors that may affect the results of studies completed with older subjects. Consequently, younger subjects may display more delays because compensation has not yet occurred (Bruyer et al., 1985, Field et al., 1978). Completely asymptomatic acallosals are rare; those who are identified by chance tend to demonstrate very mild behavioral signs (Bruyer et al., 1985), or had been thought to have had general developmental delays as children (Field et al., 1978).

Cognitive development

Measured intelligence levels of persons with agenesis of the corpus callosum tend to range from mild retardation to the average range. Chiarello (1980) reviewed 29 cases. Of those cases only one could be considered mildly retarded using AAMR standards (American Association on Mental Deficiency, 1983). Ten cases fell in the low average (borderline) range of intellectual functioning, and sixteen were in the average range.
Another review was completed by Field and her colleagues (1978). All but eight cases appear to duplicate Chiarello (1980). Of those eight, four were mildly retarded, and the remainder were functioning at higher levels. Hynd and Willis (1988) note that on average, general cognitive ability of acallosals tends to fall in the low-average range.

Lacey (1985) reviewed 40 cases of partial and total agenesis that were identified during childhood. His medical review suggests that children who are identified in early childhood, particularly those identified during infancy, are at greater risk of having mental retardation and seizures than those identified after the age of four years.

Others have hypothesized that children with callosal agenesis tend to have developmental delays for which compensation of some type occurs in addition to developmental processes (Bruyer et al., 1985; Field et al., 1978). Meerwaldt's (1983) description of an eight year old girl appears to support this hypothesis. She had been identified as acallosal at eight months of age and was described as having "retarded development and macrocephaly" (p. 161). At the age of eight, her intelligence test scores were in the average range.
The adolescent boy studied by Solursh, Margulies, Ashem and Stasiak (1965) also lends credence to this hypothesis. This subject had early intelligence test scores that placed him in the low average range of functioning. He had been assumed by his family to be mentally retarded and was reported to have been passed through some grades in school solely due to his age. When tested at age fourteen, his scores were in the average to above average range, with verbal scores one standard deviation below his performance scores.

Language development

The development of language in acallosals is of particular interest because of the lateralized nature of language functions. Dennis (1988) notes that damage in utero has widespread implications for language skill development.

In the study completed by Field and her colleagues (1978), one child was unable to describe objects, similar to symptoms shown by transsection patients. In addition, the child had an articulation problem with sound substitutions. At the time of the study, the child's sentence structure and length was adequate for her age, but one year follow-up indicated no significant development in her skills.
The infant studied by Teeter & Hynd (1981) showed general delays in language, however it appeared that normal abilities were developing. The results suggested that the developmental pattern was more similar to the Down Syndrome controls than the normal controls. The medical information on that subject suggested diffuse brain damage in addition to agenesis of the corpus callosum that may have influenced the results.

Basser (1962) provided an early hypothesis about the lateralization of speech. He noted that some children failed to develop speech after right hemisphere lesions, suggesting that both hemispheres participate in speech prior to the development of laterality. He concluded that the hemispheres have an equipotentiality to develop speech following damage to the opposite hemisphere during childhood. Gazzaniga (1970) presents a similar view. He suggests that in young children, both hemispheres develop language and other functions until the corpus callosum becomes functional beginning at age two to three.

Moscovitch (1977) considered the equipotentiality theory to be overstated. He cited evidence of lateralization of auditory processing in early infancy, and early lateralization of gesturing as indicating that lateralization is present at an early age. Further, he
notes that Basser's (1962) data also suggests lateralization. Moscovitch suggests instead that it is right hemispheric plasticity that allows for development of language functions. The data from agenesis subjects, some of whom apparently have developed bilateral speech, indicate that the potential for right hemisphere speech exists. As Moscovitch suggested, given the probable superiority of the left hemisphere, the limits of this plasticity have become evident when specific linguistic functions are studied in acallosals.

More recent studies provide further evidence for Moscovitch's position. Studies with young infants suggest that asymmetry exists as early as two months of age (Molfese & Segalowitz, 1988). Best (1988) suggests that asymmetry exists at birth, with developmental change occurring continuously at a variety of structural and functional levels.

Specific language functions were considered in depth by Dennis (1981) on one adult subject with total agenesis of the corpus callosum. This subject had no major deficits when presented with semantic or visual cues, however, when rhyming cues were presented, it was observed that her ability to access phonological relationships was limited. Her single word receptive language was within normal
limits. Expressively, she had some syntactic errors that the researcher could not directly attribute to the agenesis. Finally, some deficits were observed in verbal memory.

Jeeves and Temple (1987) reexamined Dennis' (1981) results. They concluded that both syntactic and pragmatic components of language seemed to be a problem for the subject, particularly in terms of syntactic comprehension, use, and understanding. Deficits in metalinguistic knowledge were noted, while phonological and lexical semantics were not affected.

Jeeves and Temple (1987) compared their subjects with Dennis' (1981) results, and note that the one consistent finding among the three subjects was a deficit in naming objects from rhyming cues. Other results were less consistent. Generally, there were no significant deficits in naming objects from visual cues. Two of the three subjects were impaired on the word fluency test, the third was within one standard deviation.

In a later study (Temple, Jeeves, & Vilarroya, 1989), similar results were found in a child and an adolescent. Both subjects had normal verbal intelligence levels, and both had difficulty with rhyming tasks.
Sanders' (1989) study of a six year old child also indicates problems with syntactic comprehension. Her difficulties were related to inability to understand semantic roles in some types of sentence forms, although she was able to discriminate between those sentence forms. Her problem-solving strategies on the types of forms that were difficult were similar to that of younger children, indicating that follow-up will be necessary to determine whether the problem represents a delay or a continuing deficit.

Hynd & Willis (1988) note that in addition to the syntactic-pragmatic difficulties, persons with agenesis also tend to have difficulty suppressing ipsilateral stimuli when presented with language processing tasks.

Jeeves and Temple (1987) suggest that current data do not support that the corpus callosum is essential for normal language development, nor do they disprove the hypothesis that the corpus callosum may play a role in lateralization. Recent data (Temple et al., 1989) strengthens their argument that the function of the corpus callosum in language lateralization may be one of "fine tuning" abilities during development. Further data presented by Lassonde, Bryden, and Demers (1990) indicate that agenesis of the corpus callosum does not result in
bilateral language centers. In fact, their acallosal subjects, while less strongly lateralized than higher IQ controls, were more strongly lateralized than controls who were IQ-matched.

Temple, Jeeves, and Vilarroya (1990) find further evidence that callosal agenesis may interfere in the development of specific language development. They looked at reading ability in two older children who had previously evidenced rhyming deficits and found that while their reading levels were as expected for their age, they had difficulty with specific phonological processing. These difficulties were not sufficient to interfere with word recognition, but suggest that the corpus callosum has a role in the establishment of an efficient phonological processing route.

Another type of specific deficit is described by Buchanan, Waterhouse and West (1980) who report one case of alexithymia. Alexithymia is a specific language deficit characterized by an inability to communicate emotions. It has been observed in corpus callosum transsection patients.

Visual functioning

Early studies of the visual functioning of agenics utilized testing procedures similar to those used in studying commissurotomy patients. The research questions
tended to focus on whether visual information transferred interhemispherically to be utilized in verbal or motor responses. It has been documented in several studies that acallosals require lengthened interhemispheric transfer time on tasks that require manual response; some studies have also found increased interhemispheric transfer time with decreased visual stimulus intensity and when a verbal response is required (Jeeves, 1984).

Gazzaniga, Bogen & Sperry (1962) note that in Gazzaniga's evaluation of an acallosal boy, he performed at the level of control children. The boy was of above average intelligence, and had hydrocephalus. Tasks included visual half-field testing with a tachistoscope, where the subject is to pick the design seen from a series of five.

Solursh and his colleagues (1965) studied a fourteen year old acallosal boy with average intelligence and behavioral problems. He was able to identify designs by pointing with either hand, but only when the stimulus was presented to the left visual field. He was accurate with simple color identification, but perseverated on one color when two colors were simultaneously presented.

Solursh and his colleagues suggest that these results support the implication of the corpus callosum in bilateral transfer of visual information. No impairment was noted on
visual paired associate learning, but rapid presentation of stimuli prompted perseveration. A competing stimulus in the opposite eye contributed to confusion and frustration in the controls during a paired associate learning task, but did not seem to bother the subject or affect his performance (Solursh et al., 1965).

Jeeves (1969) noted that both the adolescent and the adult acallosal subjects in his study were significantly slower at responding with both hands to lateralized visual stimulation than the controls. He cites this as evidence that acallosals utilize alternate pathways in the interhemispheric transmission of visual information on tasks that require motoric response.

Foxman and his colleagues (1986) question the extent to which the anterior commissure can compensate in the transfer of visual information. Behavioral testing has shown that the anterior commissure can transfer visually-related information in the monkey, but not humans with transsected corpus callosums. Their hypothesis is that a species-specific dissimilarity in hemispheric communication exists. With evolution, the increased need for commissural fibers appears to have occurred in the corpus callosum, leaving the amount of anterior commissure fibers relatively stable. Humans may utilize the anterior
commissure to transfer some information between temporal lobes, but not at a level of complexity to complete complex visual tasks.

The results obtained by Martin (1985) in an acallosal with no other neurological involvement provide evidence that visual identification information transfers contralaterally while spatial information does not.

Other results of visual tasks that have been found in at least one agenic subject include: decreased accuracy in detecting the discrepancy in chimeric half-faces; difficulty in identifying differences in dot density patterns presented to opposite visual half fields; and problems estimating midline stereoscopic depth at midline (Jeeves, 1984).

**Visual-motor integration and bimanual coordination**

Minor motor coordination deficits were noted in most of the cases studied in the literature reviewed here. Various studies have specifically addressed bilateral motor transfer, reaction times to lateralized visual stimuli, and various academic tasks such as drawing, writing, and puzzle completion.

Ettlinger (1977) notes that most studies reveal some motor and visual-motor deficits in acallosals, but that on most tasks, there are no significant differences from
controls. Ettlinger's early studies found significant impairment in crossed tactile localization, and matching of visual patterns in the left and right visual half-fields. He finds there is no satisfactory explanation of the variability, other than the effect of associated anomalies.

The review completed by Field and her colleagues (1978) of 24 cases indicated deficits in visual-motor abilities in each study reviewed. These include deficits in visual-motor and bimanual coordination, latency in crossed hemispheric reaction times, and significant discrepancies between reading and writing abilities.

In a series of studies conducted by Jeeves (1965, 1969) the results suggest that acallosals utilize alternate pathways in the interhemispheric transmission of visual information that requires motoric response. In one study Jeeves (1969) measured motoric response to light flashed in either visual field. The acallosals were significantly slower at responding than the normals. The slower reaction times were thought to be as a result of the time it took the information to reach the contralateral hemisphere via an alternate route.

Similar results were noted in a series of studies by Milner and his colleagues (Milner, Jeeves, Silver, Lines, & Wilson, 1985; Milner, 1982). They noted that crossed
reactions to simple visual stimuli (i.e., right hand response to left visual field stimulus) are markedly slower than uncrossed reactions in acallosals. Smaller, nonsignificant differences are found in normal individuals of average intelligence.

The study completed by Milner (1982), attempted to address the concern that the differences between crossed and uncrossed reaction times was due to below average intelligence rather than the lack of a corpus callosum. The study replicated previously observed crossed-uncrossed differences two to three standard deviations above that of normals, using a subject of below average intelligence. He did not, however use any controls of below-average intelligence.

In one of the procedures used, the hand used to respond to the stimuli was placed within the same visual field as the stimulus. The continuation of significant crossed-uncrossed differences suggested to the researchers that spatial relationships were not a factor in the differences in response time. It has still not been conclusively determined (Milner et al., 1985) what the slower response time means.
Recent work by Lassonde and her colleagues (1988) confirms that speed of task completion is slowed in persons with agenesis, while accuracy tends to be in the normal range. They suggest that increased ipsilateral spinothalamic connections is the most cogent explanation for the ability of agenics to complete tactile-motor tasks. They suggest that persons with agenesis (as well as persons who have had very early corpus callosum transsections) utilize these pathways to accomplish intrahemispheric coordination of bilateral information.

Jeeves and his colleagues (1988) used a test of bimanual coordination that included both visual and lack of visual input conditions. They made four general conclusions. First, whatever alternate pathways are available, there is likely to be a ceiling to the ability to utilize those pathways. Second, an intact corpus callosum is necessary for quick, coordinated bimanual performance. Third, acallosals appear to develop lateral dominance of motor control, calling into question the hypothesis that the corpus callosum plays an exclusively inhibitory role in the development of asymmetry. Fourth, their results lend support to the hypothesis that there is an inhibitory role for the corpus callosum in the control of bimanual control without visual feedback.
Bimanual transfer of learning

Bimanual transfer of learning was studied with two acallosal siblings (Sauerwein et al., 1981). They report that on the Halstead-Reitan Tactual Performance Test, the younger subject was within normal limits on initial learning (preferred hand) and transfer of learning (nonpreferred hand) while his older sister was markedly slower on both tasks (Sauerwein et al., 1981). This is similar to Chiarello (1980), who found in her literature review that 100% of subjects tested by a similar task (n=6) showed deficits in initial learning and 63% showed deficits in transfer of learning.

Results obtained by Sauerwein and her colleagues (1981) indicate that the subjects performed within the normal time limits on the bimanual task, however they were well above their age norms on localization (drawing) errors. These two acallosal subjects tended to have better control on bimanual tasks than lateralized tasks, which is contrary to much of the rest of the literature.

Generally, studies using tactual form boards or maze tasks report reduced transfer of learning of the task when compared to normals (Jeeves, 1969; Reynolds & Jeeves, 1977; Russell & Reitan, 1955; Solursh et al., 1965).
Some studies have noted deficits in bimanual tasks that require vision, including buttoning, threading beads and peg board completion (Field et al., 1978; Jeeves, 1969; Reynolds & Jeeves, 1977; Sperry, Gazzaniga & Bogen, 1969). In the study completed by Field and her colleagues (1978), neither acallosal subject was able to complete a buttoning task at the age of four.

In their study, Field and her colleagues (1978) noted that one of the children was able to manipulate pegs in the pegboard with the nondominant hand only when she was not visually attending to what she was doing. When attending to task, she refused to attempt the pegs with the nondominant hand. They conclude that visual input to both hemispheres, may compete with the kinesthetic input from the hands, which is going to separate, noncommunicating hemispheres. The result is disorganized visual-motor coordination.

**Kinesthetic functioning**

Cross-lateral identification of touches on a hand screened from view (Sauerwein et al., 1981) revealed no significant differences between acallosals and controls. When asked to verbally describe which finger had been touched, the acallosals had long delays before responding. In contrast, verbal report of shapes and objects placed in the hand showed no significant differences between the hand the object was in and ability to respond verbally.
Field and her colleagues (1978) looked at kinesthetic identification in children. One child was able to identify only one of five objects in her nondominant right hand. She identified five of five objects with her left hand, and five of five objects when the right hand was retested.

The second subject was unable to identify any objects in either hand. When able to look at the objects, all five were identified correctly. When the kinesthetic-only condition was repeated, no objects were identified in either hand.

Older subjects studied by Lassonde and her colleagues (1988) were just as accurate as normal subjects when making tactile comparisons. Their response times were considerably slower than that of controls, however.

Jeeves & Silver (1988) noted that previous evidence indicated interference between uncrossed (proximal) and crossed (distal) pathways in movement. Following reanalysis of previous data, they noted that contrary to the normal pattern of grasp, their acallosal subject's grasp remained open until the object was touched. These results are similar to those reported by Gazzaniga (1970) with transsection subjects who had better control over proximal muscles than distal muscles.

**Summary**

The literature provides information on the neurodevelopmental, etiological, and structural aspects of
agenesis of the corpus callosum. It also provides evidence of functioning levels of what is primarily an older child and adult population. The present study will provide us with an understanding of the cognitive, motor, adaptive behavior and language functioning of young, preschool children with agenesis of the corpus callosum.
Subjects and setting

To determine the ways in which congenital absence of the corpus callosum affects young children, this study used the one-group posttest-only design (Campbell & Stanley, 1966; Cook & Campbell, 1979). The children in this study are served in publicly-funded early intervention preschool programs (whether home-based or classroom-based) and are medically diagnosed as having congenital agenesis of the corpus callosum. Two subjects who were initially identified from records as having total agenesis were later found to have partial agenesis. One subject with total callosal agenesis was located who was not involved in early intervention. The medical records of this pool of potential subjects were reviewed, and diagnostic films reviewed by a pediatric neurosurgeon, to confirm the diagnosis.

The participation of all subjects was voluntary, with the parents of the subjects free to withdraw their consent and discontinue the participation of their child at any time. An attempt to control for voluntary withdrawal was made by notifying the parents in advance that in return for
### TABLE 1

SUBJECT CHARACTERISTICS

<table>
<thead>
<tr>
<th>SUBJECT</th>
<th>AGE (Year-months)</th>
<th>GENDER</th>
</tr>
</thead>
<tbody>
<tr>
<td>Case 1</td>
<td>3-5</td>
<td>F</td>
</tr>
<tr>
<td>Case 2</td>
<td>5-8</td>
<td>M</td>
</tr>
<tr>
<td>Case 3</td>
<td>5-4</td>
<td>M</td>
</tr>
<tr>
<td>Case 4</td>
<td>2-3</td>
<td>M</td>
</tr>
<tr>
<td>Case 5</td>
<td>2-8</td>
<td>F</td>
</tr>
<tr>
<td>Case 6</td>
<td>0-5</td>
<td>F</td>
</tr>
<tr>
<td>Case P7*</td>
<td>4-11</td>
<td>F</td>
</tr>
<tr>
<td>Case P8</td>
<td>2-3</td>
<td>F</td>
</tr>
</tbody>
</table>

*Case numbers including P have partial agenesis.
their child's participation in the study they would receive a written report on their child's performance. If requested, they were also provided with an abridged version of the literature review.

**Procedure**

The parents of all children were interviewed to determine background history of the child. This included any known etiological factors, and developmental milestones. The Adaptive Behavior Scale for Infants and Children was completed with the parent.

Each child's school file was reviewed to gain information from annual screenings administered at the preschool to determine their developmental progression. General medical information was obtained from the preschool files. Further medical information was obtained from medical files. If necessary, their physicians were interviewed to confirm the degree of agenesis, other existing neurological anomalies, the source of the diagnosis (i.e. CT scan, MRI), and the referring problem.

A battery of psychological instruments designed to measure intelligence level, receptive and expressive language, bimanual coordination, and visual-motor abilities was administered to each child. The age of these subjects precluded the use of many standardized neuropsychological instruments, or those designed to measure the subtle effects
of callosal agenesis on vision, transfer of learning and language.

Originally, nine children between the ages of 5 months and 5 years, 8 months were evaluated. Upon closer examination of the CT and MRI films, it was found that two children who had medical records that indicated agenesis of the corpus callosum had only partial agenesis. These two children were kept in the study for post hoc analysis. They were matched by age and overall developmental level and compared with two subjects with total agenesis. A third child was found to have an intact corpus callosum, and those data were dropped from the study.

**Measures**

**Adaptive Behavior**

**Adaptive Behavior Scale for Infants and Early Childhood (ABSI):** The ABSI provides a behaviorally based indicator of a child's adaptive and maladaptive behaviors. (Leland, Shoae, McElwain, & Christie, 1980/1990; Ritchie, Simpson, Thompson, DeRienzo, Han, & Harris, 1978). This criterion-based scale is part of the standards used by the American Association on Mental Retardation for the diagnosis of mental retardation (American Association on Mental Retardation, 1983), to determine level of adaptive behavior, and adaptive strengths and weaknesses.
Cognitive and Developmental

Bayley Scales of Infant Development: The purpose of the Bayley is to measure cognitive and motor development of children who are 2 1/2 years and younger (Bayley, 1969). It is a well designed tool for the measurement of patterns of early development (Franzen & Berg, 1989; Sattler, 1988). This measure was used with acallosal subjects younger than 2 1/2 and any acallosal subject who did not have sufficient language or motor skills to score on the McCarthy scales.

McCarthy Scales of Children's Abilities: The McCarthy (McCarthy, 1972) is an individually administered test of cognitive ability suitable for children aged 2 1/2 to 8 1/2. Sattler (1988) notes that this instrument has features that make it suitable for young children with developmental delays. It provides an overall index of cognitive level, as well as an index of language, perceptual-motor, memory and gross motor functioning.

Early Intervention Developmental Profile (EIDP): The EIDP is a developmental screening tool administered annually at the preschool. It provides a general measure of a child's developmental level in six areas including cognition, fine and gross motor abilities, language, social-emotional, and self-care (Ritchie et al., 1978). Protocols from this instrument, when available, were used to supplement the developmental history of the children.
Language Development

Peabody Picture Vocabulary Test-Revised (PPVT-R): The PPVT-R (Dunn & Dunn, 1981) is a measure of one-word receptive language for which no verbalization is required, which can be adapted in case of physical handicap (Franzen & Berg, 1989; Ritchie et al., 1978; Sattler, 1988).

Preschool Language Scale (PLS): The PLS is designed to measure expressive and receptive language in preschool children from the ages of 1 through 7. It provides evidence of contextual auditory comprehension and verbal ability. (Zimmerman, Steiner & Pond, 1979).

Visual-motor and Bimanual Developmental Test of Visual-Motor Integration (DVMI): The purpose of the DVMI (Beery, 1989) is to assess integration of visual and motor skills in children. Children are asked to copy a variety of geometric forms that are arranged in order of difficulty (Franzen & Berg, 1989; Sattler, 1988).

Merrill-Palmer Scale: The Merrill-Palmer (Stutsman, 1949) is a developmental screening tool for preschool children. While it is no longer widely used, selected subtests were administered, similar to those administered by Field and her colleagues (1978) and Jeeves (1965). These subtests include the Wallin Pegs, and the buttoning task to provide measures of bimanual ability.
The Wallin Pegs were administered according to standard procedure, with two timed trials for each of the two pegboards.

The Buttoning task was administered according to standard procedure. An attempt was made to include a no-vision condition, using a curtain to screen the child visually from their performance to test the observations made informally by Field and her colleagues (1978). Attempts to administer the screened condition were rejected by all subjects, and will be reported only in anecdotal form.

**Seguin-Goddard Formboard: Arthur Adaptation:** This formboard is the forerunner of the Tactual Performance Test used with young children in the Reitan Indiana Battery (Franzen & Berg, 1989; Hynd, 1988). The Arthur adaptation of the Sequin was chosen rather than the Reitan adaptation due to the age of the subjects. The availability of norms, the raised design of the blocks, and the lack of blindfolding are more suited to preschool aged children.

**Statistical Analyses**

Due to the loss of subjects, and homogeneity of age and functioning level that precluded matching as originally proposed, nonparametric statistical comparison was not possible. All children have a case analysis of background history, medical status and results of evaluation.
Comparisons were made by means of tables, graphs and scatterplots.

Qualitative information that was observed is reported on a case by case basis in Chapter V. This includes apparent attempts by the subjects to compensate, descriptions of performance, as well as case history information.
Chapter IV
Data Analysis

Overview of the Chapter

The analysis for this chapter consists of three sections. The first section presents data obtained from medical files and background history, including associated anomalies and birth history. These data will begin to address the other factors that may affect the functioning levels of the children in this sample, in addition to their absence of a corpus callosum.

The second section analyzes current functioning levels in cognition, adaptive behavior, language, and visual-motor coordination. These analyses will address the following research questions: Will children with absence of the corpus callosum have an identifiable, specific pattern of developmental delays. Will acallosal children display delays in visual-motor tasks and language. Are there patterns of developmental delays.

The third section will address the following question: Will younger children be more likely to exhibit identifiable delays than older children with agenesis.

56
In addition, two cases will be compared with similar-aged children with partial agenesis of the corpus callosum (Subjects P7 and P8).

Background Data

One question that has been difficult to address with older acallosal subjects is the extent to which early developmental and medical factors may have contributed to variations in functioning levels. One advantage of the age of the current subjects is that retrospective data from medical files and parent recollection remains relatively intact.

Table 2 identifies the age of initial diagnosis, the means of diagnosis and symptoms prompting the diagnosis. Two subjects were identified for further diagnosis through prenatal ultrasound. The shape of their ventricles suggested hydrocephalus or ventricularmegaly. One subject was identified in early infancy due to congenital hydrocephalus. Three other subjects were identified between the ages of 1 and 3 years of age. Two cases had unusual eye shapes and head control problems that concerned their parents. The third was identified after a fall which resulted in a slight skull fracture.
<table>
<thead>
<tr>
<th>Subject</th>
<th>Age at Initial Diagnosis</th>
<th>Means of Initial Diagnosis</th>
<th>Means of Final Diagnosis</th>
<th>Symptoms Prompting Evaluation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Case 1</td>
<td>1-0</td>
<td>CT</td>
<td>MRI</td>
<td>Head lag to left, left eye drooped hit self in head with left hand.</td>
</tr>
<tr>
<td>Case 2</td>
<td>3-0</td>
<td>CT</td>
<td>MRI</td>
<td>Shook head side to side, motor delayed, eyes &quot;looked funny&quot;.</td>
</tr>
<tr>
<td>Case 3</td>
<td>0-1</td>
<td>Postnatal ultrasound</td>
<td>CT</td>
<td>Congenital hydrocephalus, dismorphic features.</td>
</tr>
<tr>
<td>Case 4</td>
<td>Prenatal</td>
<td>Amniocentesis Ultrasound</td>
<td>CT, MRI</td>
<td>Amnio: Trisomy 8p. Ultrasound: Possible hydrocephalus.</td>
</tr>
<tr>
<td>Case 5</td>
<td>2-6</td>
<td>X-ray</td>
<td>CT, MRI</td>
<td>Fell: skull fracture in left parietal area.</td>
</tr>
<tr>
<td>Case 6</td>
<td>Prenatal</td>
<td>Ultrasound</td>
<td>CT, MRI</td>
<td>Ultrasound: Possible ventricularmegaly.</td>
</tr>
<tr>
<td>Case P7</td>
<td>0-2</td>
<td>CT</td>
<td>MRI</td>
<td>Congenital hydrocephalus.</td>
</tr>
<tr>
<td>Case P8</td>
<td>Prenatal</td>
<td>Ultrasound</td>
<td>CT, MRI</td>
<td>Congenital hydrocephalus.</td>
</tr>
</tbody>
</table>
Birth complications are outlined in Table 3. As can be seen, most of the subjects had some birth complications. Perinatal hypoxia and postnatal anoxia are suggested in a number of cases. Three infants were ventilated, all of whom had some degree of fetal distress documented in their medical files. All three spent at least some time in neonatal intensive care units. The other three cases had no documented complications and remained in the hospital for a typical length of time.

The literature indicates it is not uncommon for seizure activity to be found in acallosals. Table 4 outlines seizure diagnoses and EEG results in this sample. Three subjects have exhibited seizure activity. Case 6, the 5 month old infant, was placed on a course of ACTH during the study, and continues to have infantile spasms on a daily basis, although reduced in frequency.

Case 3 had one seizure at the age of 5, and while the seizure type was not specified, he was released from the hospital with a diagnosis of a viral infection, suggesting that the seizure was febrile in nature. Case 4 had seizures at one day of age, but none have been observed since that time. His most recent EEG results indicated patterns that were normal for his age.

One case who has not exhibited seizure activity had an abnormal EEG reading. Case 1 was found to have
<table>
<thead>
<tr>
<th>Subject</th>
<th>Weeks Gestation</th>
<th>Apgars</th>
<th>Ventilated</th>
<th>Fetal Distress</th>
<th>No. Days NICU</th>
<th>No. Days Hospital</th>
<th>Delivery</th>
</tr>
</thead>
<tbody>
<tr>
<td>Case 1</td>
<td>FT</td>
<td>N.Sp.*</td>
<td>Yes</td>
<td>Yes</td>
<td>3</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Case 2</td>
<td>FT</td>
<td>N.Sp.</td>
<td>No</td>
<td>No</td>
<td>0</td>
<td>2</td>
<td>Vaginal</td>
</tr>
<tr>
<td>Case 3</td>
<td>37</td>
<td>6, 8</td>
<td>Yes</td>
<td>Low H.R.</td>
<td>35</td>
<td>42</td>
<td>Vaginal</td>
</tr>
<tr>
<td>Case 4</td>
<td>41</td>
<td>1, 1</td>
<td>Yes</td>
<td>Yes</td>
<td>5</td>
<td>15</td>
<td>Vaginal</td>
</tr>
<tr>
<td>Case 5</td>
<td>40</td>
<td>8, 9</td>
<td>No</td>
<td>No</td>
<td>0</td>
<td>5</td>
<td>Repeat C-section</td>
</tr>
<tr>
<td>Case 6</td>
<td>FT</td>
<td>8, 9</td>
<td>No</td>
<td>No</td>
<td>0</td>
<td>2</td>
<td>Vaginal</td>
</tr>
<tr>
<td>Case P7</td>
<td>FT</td>
<td>1, 4</td>
<td>Yes</td>
<td>Yes</td>
<td>N.Sp.</td>
<td>54</td>
<td>C-section</td>
</tr>
<tr>
<td>Case P8</td>
<td>39</td>
<td>N.Sp.</td>
<td>Yes</td>
<td>No</td>
<td>N.Sp.</td>
<td>21</td>
<td>C-section</td>
</tr>
</tbody>
</table>

* N.Sp.: Not specified in the records.
<table>
<thead>
<tr>
<th>Subject</th>
<th>Seizure Type</th>
<th>Age at Onset</th>
<th>EEG Results</th>
<th>Current Medication</th>
</tr>
</thead>
<tbody>
<tr>
<td>Case 1</td>
<td>N/A</td>
<td>N/A</td>
<td>Abnormal.</td>
<td>None</td>
</tr>
<tr>
<td>Case 2</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Case 3</td>
<td>N.Sp.* Febrile?</td>
<td>5-0</td>
<td>Waking: Bilateral slowing. Sleep: left hemisphere spike and wave.</td>
<td>None</td>
</tr>
<tr>
<td>Case 4</td>
<td>N.Sp.</td>
<td>Day 1</td>
<td>Normal for age.</td>
<td>None</td>
</tr>
<tr>
<td>Case 5</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Case 6</td>
<td>Infantile spasms</td>
<td>0-2</td>
<td>Waking: Bilateral slowing. Sleep: Multifocal discharge left hemisphere.</td>
<td>ACTH</td>
</tr>
<tr>
<td>Case P7</td>
<td>Partial complex</td>
<td>3-11</td>
<td>Slow wave:Right hemisphere.</td>
<td>Phenobarbital</td>
</tr>
<tr>
<td>Case P8</td>
<td>N/A</td>
<td>N/A</td>
<td>Waking: movement artifact. Sleep: multifocal discharges.</td>
<td>Mellaril**</td>
</tr>
</tbody>
</table>

* N.Sp.: Not specified

** Prescribed for agitation and self-injurious behavior unrelated to EEG findings.
independent bilateral foci in the occipital region, with
general right hemisphere slowing. She is not currently
prescribed any medications.

Anomalies noted in the medical records are presented
on Table 5. The only chromosomal anomaly in this sample is
Case 4, who has a diagnosis of Trisomy 8p.

Craniofacial irregularities are also noted. They
include asymmetrical and dysmorphic features,
irregularities of jaw placement or size, and one case with
bilateral epicanthal folds noted in the records.

No consistency or trend was noted in the diagnosis of
craniofacial anomalies. However, observation of the
subjects indicated facial similarities that bear noting.
All the children had some degree of hypertelorism,
epicanthal folds or the appearance of folds, and a
similarity of shape due to jaw or bite configuration. This
resulted in a strong resemblance across the subjects. The
one exception was Case 6, however the edema resulting from
the ACTH treatment affected her facial shape, and age is
likely to play a factor.

Neurological anomalies include one case with
hydrocephalus and one case with porencephaly. Two cases
have dilation and displacement of the lateral and third
ventricles noted in their medical records. Three cases have
<table>
<thead>
<tr>
<th>Subject</th>
<th>Chromosomal</th>
<th>Neurological</th>
<th>Craniofacial</th>
<th>Other</th>
</tr>
</thead>
<tbody>
<tr>
<td>Case 1</td>
<td>None</td>
<td>Total agenesis of CC, optic nerve hypoplasia.</td>
<td>Deep bite, extra tooth at midline.</td>
<td>Bilateral space between forefinger and second finger, frenulum tie laryngomalacia.</td>
</tr>
<tr>
<td>Case 2</td>
<td>None</td>
<td>Total agenesis of CC, left optic nerve hypoplasia.</td>
<td>Small maxilla.</td>
<td>Bilateral clinodactyly of 5th finger, frenulum tie, left exotropia.</td>
</tr>
<tr>
<td>Case 3</td>
<td>None</td>
<td>Total agenesis of CC, hydrocephalus: not shunted, dilation of left posterior horn and 3rd ventricles.</td>
<td>Mild retrognathia, prominent forehead, bilateral epicanthal folds.</td>
<td>Club foot, umbilical hernia bilateral inguinal hernia, mild cardiomegaly, alternating esotropia.</td>
</tr>
<tr>
<td>Case 4</td>
<td>Trisomy 8p</td>
<td>Total agenesis of CC, incomplete myelination, micrognathia.</td>
<td>Dysmorphic features, parallel wide-set lateral ventricles, high 3rd ventricle.</td>
<td>Failure to thrive, cardiac anomalies, submucous cleft, left esotropia, chalasia bilateral inguinal hernia coarctation of aorta, ventral septal defect, hypospadius.</td>
</tr>
<tr>
<td>Case 5</td>
<td>None</td>
<td>Total agenesis of CC.</td>
<td></td>
<td>None</td>
</tr>
<tr>
<td>Case 6</td>
<td>None</td>
<td>Total agenesis of CC, Porencephaly: Left frontal pole.</td>
<td>Asymmetrical features.</td>
<td>Microthalamic left eye with coloboma, right cortical blindness, hemangiomas on left foot and left nipple.</td>
</tr>
<tr>
<td>Case P7</td>
<td>None</td>
<td>Partial agenesis of CC, hydrocephalus: VP shunt arachnoid cyst, dilation of lateral ventricles.</td>
<td></td>
<td>Alternating esotropia, hip adduction.</td>
</tr>
<tr>
<td>Case P8</td>
<td>None</td>
<td>Partial agenesis of CC, Craniostenosis. Hydrocephalus: VP shunt.</td>
<td></td>
<td>Failure to thrive, reflux bilateral esotropia.</td>
</tr>
</tbody>
</table>
visual anomalies. Two have hypoplastic optic nerves, and one has a diagnosis of cortical blindness, microphthalmia and coloboma. The two cases with partial agenesis both had hydrocephalus, one with craniostenosis (Case P7) and the other with an arachnoid cyst (Case P8).

Other anomalies include esotropia and two cases with cardiac irregularities. Medically, three subjects have a history of frequent upper respiratory infections, two have asthma, and three have a history of frequent ear infections.

Current functioning

Three cases were administered the McCarthy Scales of Children's abilities as a measure of cognitive functioning (see Table 6). Two cases under the age of 30 months were administered the Bayley Scales (see Table 7). Cases 3 and P7, although older, were administered the Bayley Scales due to their severe levels of developmental delays.

Two cases have measured intelligence levels in the moderate deficit range or below. One case is in the mild deficit range. Two cases are currently functioning in the low average range, and one case is in the average range.

Figure 1 indicates the pattern of results obtained on the McCarthy Scales. The only pattern that appears to emerge is that given verbal and perceptual-performance
<table>
<thead>
<tr>
<th>Subject</th>
<th>C.A. (x=100, sd=16)</th>
<th>General Cognitive Index</th>
<th>Verbal</th>
<th>Perceptual-Performance</th>
<th>Quantitative</th>
<th>Memory</th>
<th>Motor</th>
</tr>
</thead>
<tbody>
<tr>
<td>Case 1</td>
<td>3-5</td>
<td>81</td>
<td>41</td>
<td>32</td>
<td>42</td>
<td>44</td>
<td>37</td>
</tr>
<tr>
<td>Case 2</td>
<td>5-8</td>
<td>50</td>
<td>30</td>
<td>24</td>
<td>23</td>
<td>23</td>
<td>31</td>
</tr>
<tr>
<td>Case 5</td>
<td>2-8</td>
<td>102</td>
<td>46</td>
<td>60</td>
<td>56</td>
<td>52</td>
<td>54</td>
</tr>
<tr>
<td>Subject</td>
<td>C.A. (Months)</td>
<td>Mental Development Index</td>
<td>MDI Age Equivalent</td>
<td>Psychomotor Development Index</td>
<td>PDI Age Equivalent</td>
<td></td>
<td></td>
</tr>
<tr>
<td>---------</td>
<td>---------------</td>
<td>--------------------------</td>
<td>-------------------</td>
<td>-----------------------------</td>
<td>-------------------</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Case 3</td>
<td>5-2</td>
<td>N/A</td>
<td>9-12 mo.</td>
<td>N/A</td>
<td>9-11 mo.</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>(62 mo.)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Case 4</td>
<td>2-3</td>
<td>50</td>
<td>12-14 mo.</td>
<td>50</td>
<td>9-10 mo.</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>(27 mo.)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Case 6</td>
<td>0-5</td>
<td>85</td>
<td>4-4.5 mo.</td>
<td>85</td>
<td>4-4.5 mo.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Case P7</td>
<td>4-11</td>
<td>N/A</td>
<td>15-17 mo.</td>
<td>N/A</td>
<td>17-19 mo.</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>(59 mo.)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Case P8</td>
<td>2-3</td>
<td>50</td>
<td>6-10 mo.</td>
<td>50</td>
<td>6-10 mo.</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>(27 mo.)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
FIGURE 1

MCCARTHY RESULTS
### TABLE 8

**ADAPTIVE BEHAVIOR RESULTS**

<table>
<thead>
<tr>
<th>Subject</th>
<th>AAMR Level*</th>
<th>IF**</th>
<th>PF</th>
<th>Communication Skills Percentiles</th>
<th>Concept Skills</th>
</tr>
</thead>
<tbody>
<tr>
<td>Case 1</td>
<td>1</td>
<td>82</td>
<td>80</td>
<td>68</td>
<td>25</td>
</tr>
<tr>
<td>Case 2</td>
<td>1</td>
<td>80</td>
<td>62</td>
<td>99</td>
<td>15</td>
</tr>
<tr>
<td>Case 3</td>
<td>2-3</td>
<td>12</td>
<td>28</td>
<td>32</td>
<td>30</td>
</tr>
<tr>
<td>Case 4</td>
<td>2-3</td>
<td>65</td>
<td>78</td>
<td>62</td>
<td>72</td>
</tr>
<tr>
<td>Case 5</td>
<td>0</td>
<td>85</td>
<td>90</td>
<td>38</td>
<td>40</td>
</tr>
<tr>
<td>Case 6</td>
<td>0</td>
<td>98</td>
<td>0</td>
<td>92</td>
<td>99</td>
</tr>
<tr>
<td>Case P7</td>
<td>2-3</td>
<td>32</td>
<td>45</td>
<td>62</td>
<td>60</td>
</tr>
<tr>
<td>Case P8</td>
<td>2-3</td>
<td>12</td>
<td>24</td>
<td>20</td>
<td>50</td>
</tr>
</tbody>
</table>

* Adaptive Behavior Levels: 0=No significant deficits 1=Mild deficits 2-3=Moderate to Severe deficits

** IF=Independent Functioning  PF=Physical Functioning
skills, relative strength in one area is offset by relative weakness in the other. Children who were administered the Bayley scales tended to have similar levels of cognitive and motor skills.

Table 8 indicates adaptive behavior levels from the ABSI. Two cases are currently exhibiting moderate to severe deficits, two cases are exhibiting mild deficits, and two cases currently have no significant deficits. Graphing the percentile scores (Figs. 2 and 3) indicates there are two groups with similarities in relative strengths and weaknesses.

One group (Cases 3, and 4) had adaptive levels in the moderate to severe deficit range and cognitive levels in the moderate range. Independent Functioning and Physical Functioning are relative weaknesses, with relative strengths in Self Direction and Social skills.

The second group (Cases 1, 2, and 5) all had adaptive levels in the no significant deficit to mild deficit range. Cognitive levels were in the low average to average range, except for Case 2 in the mild deficit range. Case 6 was omitted from the figures due to her age, but her profile most resembled the second group.

Table 9 indicates the results of the verbal measures. Generally, all subjects obtained age equivalents lower than expected for age. The one exception was Case 5 who scored
*IF: Independent functioning; PF: Physical functioning; Comm: Communication; Con: Concept skills; Play: Play skills; SDir: Self Direction; PS: Personal/social skills.

FIGURE 2

ABSI RESULTS: MODERATE TO SEVERE DELAYS
FIGURE 3

ABSI RESULTS: NO SIGNIFICANT TO MILD DEFICITS
### TABLE 9

**VERBAL MEASURES**

<table>
<thead>
<tr>
<th>Subject</th>
<th>C-A</th>
<th>PPVT-R Standard Score (A-E)</th>
<th>PLS Verbal Ability A-E</th>
<th>PLS Auditory Comprehension A-E</th>
</tr>
</thead>
<tbody>
<tr>
<td>Case 1</td>
<td>3-5</td>
<td>78 (2-6)</td>
<td>2-10</td>
<td>2-10</td>
</tr>
<tr>
<td>Case 2</td>
<td>5-8</td>
<td>67 (3-7)</td>
<td>4-0</td>
<td>3-10</td>
</tr>
<tr>
<td>Case 3</td>
<td>5-4</td>
<td>N/A</td>
<td>1-1</td>
<td>1-1</td>
</tr>
<tr>
<td>Case 4</td>
<td>2-3</td>
<td>N/A</td>
<td>1-3</td>
<td>1-1</td>
</tr>
<tr>
<td>Case 5</td>
<td>2-8</td>
<td>90 (2-3)</td>
<td>3-3</td>
<td>3-4</td>
</tr>
<tr>
<td>Case 6</td>
<td>0-5</td>
<td>N/A</td>
<td>N/A</td>
<td>N/A</td>
</tr>
<tr>
<td>Case P7</td>
<td>4-11</td>
<td>N/A</td>
<td>1-9</td>
<td>1-1</td>
</tr>
<tr>
<td>Case P8</td>
<td>2-3</td>
<td>N/A</td>
<td>1-1</td>
<td>1-1</td>
</tr>
<tr>
<td>------------------------</td>
<td>---------------------------</td>
<td>--------------------------</td>
<td>--------------------------</td>
<td>----------------------------</td>
</tr>
<tr>
<td>Case 1 (41 mo.)</td>
<td>N/A (33 mo.)</td>
<td>21 sec. (24-29 mo.)</td>
<td>24 sec. (30-35 mo.)</td>
<td>240 sec. (30 mo.)</td>
</tr>
<tr>
<td>Case 2 (68 mo.)</td>
<td>66 (42 mo.)</td>
<td>17 sec. (36-41 mo.)</td>
<td>19 sec. (36-41 mo.)</td>
<td>147 sec. (30-35 mo.)</td>
</tr>
<tr>
<td>Case 5 (32 mo.)</td>
<td>N/A (38 mo.)</td>
<td>26 sec. (18-23 mo.)</td>
<td>23 sec. (36-41 mo.)</td>
<td>120 sec. (30-35 mo.)</td>
</tr>
</tbody>
</table>
higher than age on the PLS. Her PPVT-R standard score and age equivalent was lower than expected when scores obtained on other measures are considered. All subjects who were administered both verbal measures scored lower on the PPVT-R than on the PLS.

Table 10 indicates results on the visual-motor instruments administered to the three cases were able to complete the tasks. Two cases had scores that indicate delays in this area, while one case had scores at or near age level.

Case 1 had delays ranging from 6 to 12 months below age. She had stopped responding near the end of a second trial on the Seguin formboard with a time closer to age, but refusals resulted in an incomplete trial. She performed the tasks in a relatively random manner, skipping holes in the Wallin pegboard then going back to complete the board. Holes to the left of midline tended to be completed with the left hand, and those to the right of midline with the right hand. See Figure 4 for Wallin results.

Case 2 had delays ranging from 26 to 33 months below age. He tended to respond slowly and methodically to all tasks, and made attempts at task avoidance by asking questions. On the Wallin pegboard, he consistently skipped the second hole, completed the remainder, then went back. He completed all the pegs with his right hand.
Case 1: Dominance not established

Case 2: Dominance not established

Case 5: Dominance not established

FIGURE 4

PATTERNS OF COMPLETION: WALLIN PEGBOARD
Case 5 completed all instruments at or above age level, with the exception of Pegboard A. She was somewhat distracted during the completion of that pegboard, which may have contributed to her slower than expected performance. On the Pegboards, all pegs to the left of midline were completed with the left hand, and those to the right with the right hand.

All three cases who completed visual motor tasks were also presented with the Stutsman Buttoning task. None of the cases could button the one button task. All gave up very quickly. They seemed to have no strategy for completing the task and were not assisted by demonstration. Descriptions of individual performances can be found in the chapter on case studies.

Developmental trends

Previous research suggests that as acallosal children develop, they tend to exhibit higher levels of functioning. While this question is best answered through longitudinal study, the range of ages in the current study lends itself to discussion of functioning level and age. Additionally, the presence of background developmental data on three subjects allows for some longitudinal intrasubject comparisons.

Figure 5 is a scatterplot comparing age and measured intelligence level. As can be seen, the older children in
*AAMR ranges used: NSD=No significant deficits; MILD=Mild deficits; MOD/SEV= Moderate to severe deficits

FIGURE 5

SCATTERPLOT: COMPARISON OF AGE AND MEASURED INTELLIGENCE LEVELS
Figure 6

Scatterplot: Comparison of Measured Intelligence and Number of Complications
Adaptive Behavior Levels

*AAMR ranges used: NSD=No significant deficits; Mild=Mild deficits; MOD/SEV= Moderate to severe deficits.

FIGURE 7

SCATTERPLOT:
COMPARISON OF ADAPTIVE BEHAVIOR LEVELS
AND NUMBER OF COMPLICATING FACTORS
the sample tended to have lower levels of measured intelligence. An alternative hypothesis is that other developmental and medical factors may have affected functioning levels. Figure 6 is a scatterplot that compares measured intelligence levels and number of complicating factors. Figure 7 provides an alternate comparison of adaptive behavior level and number of complicating factors. The factors considered were: chromosomal anomaly, hydrocephalus, seizure activity, and need for ventilation at birth. A stronger relationship exists between overall functioning level and number of complicating factors than between measured intelligence level and age.

Case 1 is the only subject for which there is previous standard score equivalent data. She was administered the Bayley at the age of 12 months. Comparison of her current McCarthy GCI and the Bayley MDI can be seen on Figure 8. For this case, her measured intelligence level currently is significantly closer to the mean than during infancy. This may be an artifact of maturation, comparing different instruments, regression toward the mean, or a reflection of poor test-retest reliability in young children. The results suggest that some compensation may have occurred over time.

Figure 9 indicates previous age-equivalent scores from the Early Intervention Developmental Profile (EIDP)
FIGURE 8
CHANGE IN MEASURED INTELLIGENCE LEVEL OVER TIME:
CASE 1

* Bayley Scales MDI
**McCarthy Scales GCI
FIGURE 9

CASE 1: COMPARISON OF DEVELOPMENTAL LEVELS ACROSS DOMAINS

*Mean EIDP age equivalents.
**Mean of current evaluation instruments.
for Case 1, administered at 12 months and again at 39 months. These age-equivalents provide an estimate of functioning level over time. It appears that Case 1 has made slower than normal, but relatively steady progression across developmental domains. Also graphed are mean age-equivalent scores in fine-motor and language from the current evaluation. The data suggest a fairly high rate of development over a two month time period, but may be an artifact of comparing age equivalents from different norm groups.

Only previous age-equivalent scores are available for Case 2 and Case 3. Data from the EIDP for Case 2 is presented in Figure 10 and Table 11. Age-equivalent scores are available at 31 months and at 54 months. Over the 15 month time period, Case 2 made progress in gross motor and cognitive skills, little progress in language skills, and relatively rapid progress in fine motor skills. Comparison with current perceptual-motor age-equivalents suggests that in the past 14 months, his development in this area has leveled off somewhat. In contrast, comparison of current language functioning suggests that his language skills have shown a relatively rapid increase over the same period.

Developmental data for Case 3 are presented in Figure 11 and Table 12. In contrast to the previous two cases, Case 3 has made little progress over the past four years.
**FIGURE 10**

**CASE 2: COMPARISON OF DEVELOPMENTAL LEVELS ACROSS DOMAINS**

*Mean EIDP age equivalents.
**Mean of current evaluation instruments.
### TABLE 11
**CASE 2: PREVIOUS DEVELOPMENTAL LEVELS**

<table>
<thead>
<tr>
<th>C. A.</th>
<th>Gross Motor</th>
<th>Fine Motor</th>
<th>Cognition</th>
<th>Language</th>
</tr>
</thead>
<tbody>
<tr>
<td>31 mo.*</td>
<td>27 mo.</td>
<td>15 mo.</td>
<td>21 mo.</td>
<td>21 mo.</td>
</tr>
<tr>
<td>54 mo.</td>
<td>35 mo.</td>
<td>42 mo.</td>
<td>31 mo.</td>
<td>23 mo.</td>
</tr>
<tr>
<td>68 mos.</td>
<td>42 mo.**</td>
<td></td>
<td></td>
<td>46 mo.***</td>
</tr>
</tbody>
</table>

* Mean EIDP Age equivalent scores

** DVMI Age equivalent

*** Mean of PLS and PPVT-R scores
FIGURE 11

CASE 3: COMPARISON OF DEVELOPMENTAL LEVELS ACROSS DOMAINS

* Mean EIDP age equivalents.
** Mean of current evaluation instruments.
**TABLE 12**

**CASE 3: PREVIOUS DEVELOPMENTAL LEVELS**

<table>
<thead>
<tr>
<th>C. A.</th>
<th>Gross Motor</th>
<th>Fine Motor</th>
<th>Cognition</th>
<th>Language</th>
</tr>
</thead>
<tbody>
<tr>
<td>11 mo.*</td>
<td>4 mo.</td>
<td>4 mo.</td>
<td>4 mo.</td>
<td>7 mo.</td>
</tr>
<tr>
<td>23 mo.</td>
<td>7 mo.</td>
<td>8 mo.</td>
<td>7 mo.</td>
<td>7 mo.</td>
</tr>
<tr>
<td>64 mos.</td>
<td>10 mo.**</td>
<td></td>
<td></td>
<td>12 mo.***</td>
</tr>
</tbody>
</table>

* Mean EIDP age equivalent scores
** Mean Bayley PDI age equivalent
*** Mean Bayley MDI age equivalent
The EIDP administered at 11 months and 23 months indicates age-equivalent levels similar to those obtained on the Bayley in the current data at the age of 5 years, 4 months.

The data presented here suggest a relationship between age, associated anomalies, and rate of development. Acallosal children who have relatively fewer complicating factors tend to be higher functioning, and have had a relatively steady rate of development. Children with relatively more complicating factors tend to be lower functioning and may have a significantly slower rate of development.

**Partial agenesis of the corpus callosum**

Two subjects, Case P7 and Case P8 were found to have only partial agenesis of the corpus callosum when their MRI films were examined. Due to similarities of age and functioning level, Case P7 was matched with Case 3 and Case P8 was matched with Case 4 for purposes of comparison.

As can be seen on Table 3, all four children had significant birth histories and lengthy neonatal hospital stays. Data reported on Table 2 indicates that Case 3, Case P7, and Case P8 have hydrocephalus. Cases P7 and P8 are shunted, while Case 3 is not. Case 4's initial diagnosis suggested hydrocephalus due to the degree of ventricular dilation. His final diagnosis is Trisomy 8P. All four
TABLE 13

COMPARISON OF FUNCTIONING LEVELS:
SUBJECTS WITH AGENESIS AND PARTIAL AGENESIS
MATCHED BY AGE

<table>
<thead>
<tr>
<th>Subject</th>
<th>C.A. (Months)</th>
<th>Bayley MDI (A-E)</th>
<th>Bayley PDI (A-E)</th>
<th>ABSI Level</th>
<th>PLS Verbal Ability</th>
<th>PLS Auditory Comprehension</th>
</tr>
</thead>
<tbody>
<tr>
<td>Case 4</td>
<td>2-3 (27 mo.)</td>
<td>50 (12-14 mo.)</td>
<td>50 (9-10 mo.)</td>
<td>Moderate/ Severe</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Case P8</td>
<td>2-3 (27 mo.)</td>
<td>50 (6-10 mo.)</td>
<td>50 (6-10 mo.)</td>
<td>Moderate/ Severe</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Case 3</td>
<td>5-2 (62 mo.)</td>
<td>N/A (9-12 mo.)</td>
<td>N/A (9-11 mo.)</td>
<td>Moderate/ Severe</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Case P7</td>
<td>4-11 (59 mo.)</td>
<td>N/A (15-17 mo.)</td>
<td>N/A (17-19 mo.)</td>
<td>Moderate/ Severe</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
subjects (as noted on Table 5) have a number of additional neurological anomalies noted in their records.

Comparison of current functioning levels is on Table 13. In terms of measured or estimated intelligence levels all four subjects are functioning in the moderate deficit range or below. Standard scores could not be obtained on Case 3 or Case P8. The McCarthy should have been administered, according to their age, but neither child's developmental level was appropriate for that instrument. The McCarthy was attempted with Case P8, but she could only partially complete two items. Case 3 is neither ambulatory nor verbal. There are no significant differences between scores or age-equivalents on the Mental and Motor scales for any of the cases.

All four have significant deficits in language. Case P7 has a higher verbal ability level than the others who are completely or relatively nonverbal. They all demonstrated an understanding of a very simple question, and all echoed or imitated words or sounds. Case P7 is verbal, can make her needs known with words, combines words, and refers to herself by her name. She might have scored higher, but often refused to comply with demands.

Adaptive behavior levels are compared on Figure 12. The trend is similar across the four subjects. Independent
FIGURE 12

ABSI RESULTS: COMPARISON OF SUBJECTS WITH PARTIAL AND TOTAL AGENESIS

* IF=Independent functioning; PF=Physical functioning; Comm= Communication; Con= Concept; SDir=Self Direction; PS=Personal/social skills.
functioning is relatively weak, with strengths in self-direction.

Comparison of Case 3, with a chronological age of 5 years, 4 months, and Case P8 with a chronological age of 4 years, 11 months suggests that the child with partial agenesis is currently functioning at a higher level. She is ambulatory, but ataxic, and verbal. In contrast, Case 3 is vocal, but not verbal. He stands alone briefly and cruises on furniture, but is not currently ambulatory.

Comparison of Case 4 and Case P8, both with a chronological age of 2 years, 3 months suggests that the acallosal child is currently functioning at a similar to slightly higher level to the child with partial agenesis. Case 4 is more mobile, and uses a few words. Case P8 is able to move around on her back, but has no other independent means of mobility. She has no verbal and limited gestural and vocal communication.

The similarities between these four cases are many. All four have significant motor delays. They have significant expressive and receptive language delays, and can follow only a few simple one-step directions. They have very short attention spans and are easily distractible. None of the four cases consistently coordinate their visual and fine motor abilities, leading to limited use of objects. All four tended to reach and grasp objects without
coordinating their vision and their reach. Cases 3, P7 and P8 all were resistive of the structure imposed by the testing environment, and appeared to prefer to initiate social interactions. Case 4 was more tolerant of social interaction and intrusion by an adult.

Overall, the two cases of partial callosal agenesis do not differ significantly from the two similar-aged cases of total agenesis. All four of these cases are significantly more impaired than the other subjects. They have more significant associated anomalies, including chromosomal abnormalities and hydrocephalus. Hydrocephalus was diagnosed only in cases with significant developmental delays, and appears to be a significant predictor of functioning level.

Summary

The analyses presented in this chapter suggest that in children with agenesis of the corpus callosum, overall functioning level is related to the type and number of associated complications and anomalies present. The next chapter provides a more detailed analysis of each case.
CHAPTER V
CASE STUDIES

This chapter will provide a more inclusive analysis of each subject, including background history, previous assessments and behavior during the current evaluation.

Subjects with Agenesis of the Corpus Callosum

Case 1

Case 1 is a 3 year, 3 month old girl who was first diagnosed as having agenesis of the corpus callosum at the age of 12 months. She was observed to use 4 to 5 word sentences, ask questions, and tended to be clumsy. Her face is generally symmetrical with wide set eyes, epicanthal folds and a deep bite.

Her developmental milestones were delayed. She sat unsupported by eight months, commando crawled at 17 months, and walked at 19 months. She was babbling at 12 months, said her first word by 16 months, and was speaking in two word phrases by 25 months.

Case 1 is the product of a complicated pregnancy and delivery. Mother had X-rays at approximately two weeks
gestation and was on a variety of medications for a urinary tract condition for the remainder of the pregnancy.

Case 1 was born at term. Labor lasted 1 1/2 hours, with fetal distress and meconium staining. Birth weight was 6 lbs., 1 oz. Case 1 was in the Neonatal Intensive Care unit for three days. She received oxygen for a few hours and intravenous fluids. She had trouble maintaining her body heat, and was anecdotally described as having a large head. Neither hydrocephaly nor macrocephaly have been diagnosed.

Mother reports that Case 1 was initially screened for cystic fibrosis. She was diagnosed with laryngomalacia, or flacidity of the epiglottis, which results in noisy breathing while a young infant. She has had frequent sinus, ear, and bronchial infections.

At 10 months, Case 1 was evaluated due to her mother's concerns were that she had a head lag to the left, and when sitting tended to slump toward the left and not be able to straighten herself. Her left eye was described as droopy. At times, she would hit herself lightly in the head with her left hand, and laugh. The EEG indicated abnormal right hemisphere slowing with independent left and right occipital lobe foci. No medication was prescribed.
Case 1 was diagnosed with agenesis of the corpus callosum by CT scan at 12 months, with confirmation by MR imaging at 14 months. She has no cingulate gyrus, with some enlargement of the lateral ventricles in the occipital poles. The third and fourth ventricles, brainstem and cerebellum all appeared normal, and no gray matter heterotopias were noted. Genetic testing was normal. Initially, there was concern about the development of her optic nerve. Later examination indicated that she was seeing normally. A dental evaluation indicated that she has a deep bite, an extra tooth at midline and a high palate.

Case 1 entered early intervention preschool at the age of 15 months. She received classroom programming for one year, home-based programming for the second year, and is currently re-enrolled in classes. She is also receiving private speech and physical therapy. A language evaluation at 2 years, 4 months noted a frenulum tie.

Previous Assessments

A Bayley Scales of Infant Development administered at 12 months indicated a Mental Developmental Index of 60 and an age equivalent of 7 to 8 months, and a Psychomotor Developmental Index of 50. The Early Intervention Developmental Profile was also administered at 12 months. She was described as being very tiny and hypotonic. At that time, her gross motor skills were at the 6-8 month level,
fine motor skills at the 6-8 month level, with emerging skills at the 9-11 month level. Her language and problem solving skills were at the 9-11 month level. Her social and self-help skills were at the 9-11 month level with emerging skills at the 12-15 month level.

An ABSI completed at 13 months, with mother reporting indicated that Case 1 had overall mild delays in adaptive behavior. Physical development was a significant weakness, with concept, play and independent functioning as areas of strength. Communication, self-direction and socialization were relative weaknesses.

The EIDP was administered again at the age of 3 years, 3 months. At that time, her skills were scattering significantly. Perceptual motor skills were solid at 16 to 19 months, with skills extending to 27 months. Cognitive and language skills were at the 20 to 23 month level with some items passed up to 31 months. Gross motor skills were at the 16 to 19 month level with some skills up to 35 months.

Tests Administered

McCarthy Scale of Children's Abilities

Case 1 needed frequent reinforcement and redirection to task to maintain her cooperation. She informed the investigator that she was finished on the first item of the second subtest, and stood up to leave on the third item.
She obtained a General Cognitive Index of 81, with relative strengths in verbal, quantitative and memory skills. Perceptual-performance skills are a relative weakness.

On Block Design, she was able to stack six blocks, and partially imitated the chair and building. She tended to either stack or line up the blocks on the majority of trials, regardless of the shape she was to imitate. On Puzzle Solving, even with demonstration, Case 1 was unable to complete any of the items. Her approach to the problem was to push the two halves together. On Tapping Sequence, she used the mallet as a tool, but consistently skipped the second part of the sequence. Her grasp on the pencil was very weak. She imitated a circle, and approximated a horizontal line, but had difficulty with the vertical line. On all items she added scribbles. Her figure drawing consisted of a very light scribble followed by an attempt to discontinue testing. On conceptual grouping, she correctly identified little, big, red and yellow.

On pictorial memory, she remembered one of the items. She identified and labeled all the pictures on picture vocabulary, and gave a one-word definition for one oral vocabulary item. On verbal memory, she could repeat the last word or two of all word sequences and sentences presented. She did not remember the whole sequence on any
of the items. She was able to recall up to three digits on numerical memory. She could come up with examples of three of the categories on verbal fluency when asked, and was successful with one of the opposite analogies.

Motorically, she walked on tiptoe, walked backward for a few steps, and was partially able to walk on the straight line. She was eager to show that she could stand on one foot, but was not able to do so without holding onto something. She was able to bounce the ball one time. On the beanbag catch, she caught the bag once, but tended not to move to accommodate the throw, or moved only after the bag touched her. She hit the target throwing left handed, but could only throw the bag a few inches with her right hand. She was able to imitate all actions except thumb twiddling. When her thumbs got tangled up, she attempted to twiddle her forefingers.

**Adaptive Behavior Scale for Infants and Early Childhood (ABSI)**

The subject's mother was the informant on the ABSI. In the area of independent functioning, Case 1 is independent in feeding and drinking skills. Toilet training has begun, but she has frequent accidents. She undresses herself independently, and partially dresses, but has difficulty with fasteners. She cooperates with bathing and attempts to brush her own teeth.
In the area of physical development, she has no visual or auditory limitations. She walks but frequently falls. She walks up and down stairs without alternating feet. She can jump, but has difficulty standing or hopping on one foot.

In the communication domain, she points to familiar objects when named, consistently follows one step and inconsistently follows two step related directions. Expressively, she uses five word sentences and can be understood by strangers at least half the time. She carries on simple conversations.

Conceptually, she identifies three or more body parts, matches simple shapes, has number concepts up to two, and knows that money is used to buy things. In the area of play, she engages in exploratory play, and simple imaginative play. She enjoys physical and motor play with adults and children.

In the area of self-direction, she chooses activities, and prefers to perform tasks she can do herself. She will concentrate on a preferred activity for at least ten minutes. In the area of personal responsibility and socialization, she will tattle on others, inhibits to "no," and will stay away from dangerous objects. She comforts an unhappy person, knows the names of friends and neighbors, and likes to show others when she has done something she thinks they will like.
Language Tests

Preschool Language Scale. On the PLS, Case 1 obtained an age-equivalent of 2 years, 10 months on both the auditory comprehension and verbal ability portions. Her verbal skills scattered slightly, while her comprehension skills did not.

Case 1 can follow simple directions, and identified objects by their use. She understood big and little, but had trouble with long and short. She did not distinguish time in pictures, group like objects, or recognize colors when named. She can state her name, names a variety of objects, and repeated simple sentences. She had trouble with plurals, opposites, and with repeating longer sentences. Rote counting to ten is emerging.

Peabody Picture Vocabulary Test-Revised. On the PPVT-R, Case 1 obtained a standard score equivalent of 78 and an age-equivalent of 2 years, 6 months. Her response to the items was rapid, and she was more interested in trying to turn the page than attend to the task. She responded with her right hand, and the majority of her incorrect responses were to the right of midline.

Perceptual-Performance and Bimanual Tasks

Seguin Formboard. Case 1 took 4 minutes on the initial trial, which corresponds to an age-equivalent of below 2 years, 6 months. She tended to try to force the
TABLE 14
CASE 1: SUMMARY OF RESULTS

Chronological age: 3 years, 6 months

<table>
<thead>
<tr>
<th>McCarthy Scales</th>
<th>Scaled Score</th>
<th>Percentile</th>
</tr>
</thead>
<tbody>
<tr>
<td>General Cognitive Index</td>
<td>81</td>
<td>10</td>
</tr>
<tr>
<td>Verbal Scale Index</td>
<td>41</td>
<td>15</td>
</tr>
<tr>
<td>Perceptual/Performance Index</td>
<td>32</td>
<td>3</td>
</tr>
<tr>
<td>Quantitative</td>
<td>42</td>
<td>20</td>
</tr>
<tr>
<td>Memory</td>
<td>44</td>
<td>25</td>
</tr>
<tr>
<td>Motor</td>
<td>37</td>
<td>10</td>
</tr>
</tbody>
</table>

Preschool Language Scale
Verbal Ability
Age Equivalent 2-10
Auditory Comprehension
Age Equivalent 2-10

Peabody Picture Vocabulary Test-Revised
Standard Score 78
Age Equivalent 2-6

Developmental Test of Visual-Motor Integration
Standard Score N/A
Age Equivalent 2-9

Wallin Pegs-A
Time 21"
Age Equivalent 2-0 to 2-5

Wallin Pegs-B
Time 24"
Age Equivalent 2-6 to 2-11

Stutsman Buttoning Task
1 Button Not Completed

Seguin Formboard
Time 4' 00"
Age Equivalent 2-6
blocks into incorrect spaces. She was resistive to the second trial and gave up at 2 minutes, 43 seconds with three blocks (square, triangle and diamond) remaining.

Wallin Pegs. Case 1 completed pegboard A in 24 seconds on the first trial. The second trial was 21 seconds, which corresponds to an age equivalent of 2 years to 2 years, 5 months. Pegboard B was completed in 24 seconds on the initial trial, which corresponds to an age equivalent of 2 years, 6 months to 2 years, 11 months. The second trial took 29 seconds, because Case 1 spent time taking out the first two pegs and replacing them.

Developmental Test of Visual Motor Integration. On the DVMI, Case 1 did not copy any of the figures. She was distracted by the desire to trace her hand, rather than attend to the task.

Stutsman buttoning task. Case 1 gave up on the single button task after 34 seconds. She pushed down on the top cloth, poked it with her finger, then said "you do it." She gave up immediately when a second trial was attempted. The two button task was not administered.

Case 2

Case 2 is a 5 year, 8 month old boy. He was observed to move with slightly awkward movements, speak in 5 or more word sentences and ask a variety of questions. His face is slightly asymmetrical, with wide set eyes, slight epicanthal folds, and a recessed mandible.
When Case 2 was an infant, his mother became concerned because he tended to shake his head side to side, he didn't crawl on time, and the shape of his eyes was unusual. Final diagnosis of agenesis of the corpus callosum was at the age of three, following a CT scan.

He had a frenulectomy to correct for ankyloglossia at the age of two years, 8 months. Genetics evaluation noted no chromosomal abnormalities, but noted a small maxilla, an intact palate, and bilateral fifth finger clinodactyly. An ophthalmological evaluation indicated possible left eye exotropia, bilateral farsightedness with slightly more impairment in the left eye. He also has a hypoplastic left optic nerve.

Pregnancy and delivery were without complications. Labor lasted approximately 8 hours. As a young child, he was prone to asthma attacks, but has not had one in a year.

Early milestones were unusual. His mother reports that he rolled over at two weeks. He continued to roll over, and used rolling as a primary means of ambulation until approximately 12 months when he began to commando crawl. He walked at 12 months, and mother reports that he never really crawled in four-point. He sat up at 7 months. His mother is unsure when he said his first words. Toilet training was complete by 3 years.
Previous Assessments

Case 2 was administered the Marion Area Early Childhood assessment when he was 2 years, 7 months. At that time, his gross motor skills ranged from 24 to 30 months, his fine motor skills from 12 to 18 months, and social skills from 24 to 30 months. Cognitive, language, and self-help skills all were within the 18 to 24 month range.

He was administered the Early Intervention Developmental Profile at the age of 4 years, 6 months. At that time, his gross motor skills were at approximately 35 months, his fine motor skills were at approximately 42 months and his cognitive skills were at 31 months with some items passed up to 35 months. His language skills were at 23 months.

Tests Administered

McCarty Scales of Children's Abilities

Overall, Case 2 appeared more easily distracted and tended to use a shorter length of utterance during the evaluation than previously observed at home. He tended to be resistive of the testing structure. Overall, his General Cognitive Index was at 50, with relative strengths in verbal and gross motor skills. In general, Case 2 tended to rely on visual cues, sometimes appeared not to understand directions, and occasionally displayed motor perseveration.
Case 2 was able to imitate simple block structures, but tended to continue stacking the blocks after the initial block tower. He self-corrected enough to score on other items. On Puzzle Solving, he completed the first two items, after initially completing the first upside down then correcting it. The final puzzle attempt consisted of lining up the pieces in a row. On Tapping Sequence, he completed the initial sequence correctly, then continued to repeat that sequence on the following items. His visual attending to this task was poor after the initial item.

On the Draw-a-design subtest, Case 2 adequately imitated the examiner's designs. He had a lot of difficulty copying the designs that followed. On the Draw-a-child subtest, his person included a face with features and hair, feet and elbows. He used his left hand to draw on these subtests. On the conceptual grouping subtest, he identified size, color and shape.

On verbal subtests, his attending fluctuated. On Pictorial Memory, he correctly remembered two of six items, and gave an associated response on a third. He correctly identified all pictures on Word Knowledge. He also correctly defined two words from verbal cues only. He stopped responding after the examiner had difficulty understanding his utterance on one of the items. On verbal memory, he remembered at least one cue on the initial items.
which consist of an unrelated string of words. He stopped attending when presented with sentences to remember, and requested to go back to the room when asked to remember the salient parts of a short story. He seemed to enjoy the Verbal Fluency subtest, and in spite of some unrelated responses on the last two items, gave at least one appropriate response on all items. He was able to give responses to the initial items on Opposite Analogies with little trouble.

On Quantitative subtests, his performance seemed affected by not understanding what was required on some items. He does not consistently have number concepts beyond two. On Number Questions, he did not appear to understand several of the items. He counted on the first item, successfully completed the next two, then began giving related responses. For example, when asked a number question about toys, he responded, "I have toys at my house." He was able to repeat a series of numbers on Numerical Memory up to three digits, and did not understand what was expected on the backward digits. His performance on Counting and Sorting was very inconsistent. He was not able to take two blocks from a larger group. Instead, he continued to take blocks until the whole group was gone. When presented with a group of 5, he correctly identified "how many?.." When presented with a group of blocks, told to
put the same number on each paper, he successfully completed both tasks, but when asked "how many?" were on one paper, he was not able answer correctly.

On motor items, Case 2 was able to walk forward and backward, skipped, but lost his balance slightly on the straight line. He had more trouble with static balance; he stood on his right foot for 3 seconds, but could not stand on his left foot alone for more than two seconds. He imitated the examiner with foot, hand and eye-hand items, sighting through the tube with his right eye. He bounced the ball one time with his left hand. On catching tasks, he did not seem to understand what was expected on the first trials. He also tended to stiffen his arms in front of him to catch, and did not move his body or arms to accommodate variations in the throw. He preferred to catch the right hand, with the left the preferred hand to use for throwing.

Although hand dominance was not established according to the McCarthy criteria, he clearly tended to use his left hand more consistently than his right.

**Adaptive Behavior Scale for Infants and Early Childhood (ABSI)**

The subject's mother served as informant on the ABSI. In the area of independent functioning, Case 2 feeds and toilets independently. He is nearly independent in dressing, but has difficulty with buttons. He behaves
safely in vehicles, and can go short distances from home without getting lost.

Physically, he has no limitations in visual or auditory functioning. He walks and runs without falling, although his mother notes he went through a very clumsy stage at one point. He jumps, and hops on one foot. He does not yet hold a pencil in a mature grasp.

In the communication domain, he speaks in five or more word sentences. He uses future tense, and can usually be understood by strangers. His vocabulary includes words for action, quantity, and location. He uses pronouns, and asks a variety of questions. He converses for a variety of purposes, and will give his full name when asked.

In the area of conceptual skills, he matches and identifies a variety of shapes. He has number concepts to two. He has some size and time concepts, operates a coin machine, and knows the purpose of a watch.

In the play domain, he engages in a variety of exploratory and imaginative play activities. He assumes the role of others, uses toys in a way that corresponds to their intended use, and pretends an object is something else. He actively engages in simple group games, motor play and physical play with adults and children.
In the area of self direction, Case 2 attends to his environment, tries to take part in the activities of others, and plays with toys for an extended period. He prefers to perform activities without help if he can do it himself, and makes a variety of choices. He does not tend to initiate group activities. His attention span can last 30 minutes for an activity that he enjoys but tends to be short.

In the personal responsibility and socialization domain, he will give an excuse for something he has done wrong, tattles on others, and stays away from prohibited objects. He needs reminders to complete tasks such as cleaning up toys. If someone is upset, he will attempt to comfort them. He has information about others, and likes to show family members when he has done something that he thinks they would like.

Language Tests

**Preschool Language Scale.** On the PLS, Case 2's auditory comprehension was slightly lower than verbal ability. His comprehension age equivalent was 3 years, 10 months, and verbal age equivalent was 4 years, 0 months.

Receptively, he was able to identify pictures by their function, discriminated textures, and demonstrated time recognition. He identified colors inconsistently, grouped two sets of objects, and was able to differentiate
long and short in four different configurations. He understood some simple prepositions.

Expressively, he knew a variety of opposites, and rote counted to 10. He had trouble with the memory items on this task. He could only remember the last 2 of 6 words in a sentence, and could not repeat more than three digits.

At the one-word level, his articulation is relatively good. He passed all articulation items at the 3 year, 6 month level, and passed all but two items up to 6 years. At the sentence level, however, he can be difficult to understand.

Peabody Picture Vocabulary Test-Revised

On the PPVT-R, Case 2 obtained an age equivalent score of 3 years, 7 months, which corresponds to a Standard Score of 67. His performance was inconsistent, and characterized by a great deal of scatter. He seemed to have more difficulty identifying pictures that showed action.

Perceptual-Performance and Bimanual Tasks

Seguin Formboard. Case 2 completed the formboard in 2 minutes, 27 seconds on the first trial, and in 1 minute, 40 second on the second trial. His performance was slower than expected for his age, and is similar to children who are 30 to 35 months old. His approach to the task was slow and deliberate, with occasional off-topic conversation and distractibility that required redirection to task.
Wallin Peg boards. Case 2's performance was characterized by initial interest, with decreased attention to task on the second trial. His performance on Pegboard A was 17 seconds on the first trial, average for a child who is 36 to 41 months of age.

His performance on Pegboard B was 19 seconds on the first trial, which corresponds to an age-equivalent of 36 to 41 months of age.

Developmental Test of Visual-Motor Integration. On the DVMI, Case 2 had an emotional reaction to the task. He resisted verbally and behaviorally. He drew what he wanted rather than following directions. He tended to have a weak grasp on the pencil, and held it near the eraser. The line quality is weak. He also shifted hands, used his chin in an apparent attempt to steady the pencil, and at one point used both hands on the pencil. In spite of these difficulties, he obtained an age equivalent of 3 years-6 months and a standard score of 66. His performance on the 3-line cross was similar to that observed by Beery (1989) at the 4 year-6 month to 4 year-11 month level.

Stutsman buttoning task. The performance on the buttoning task was characterized by refusals, giving up, and lack of a consistent plan of attack. On the one-button task, he gave up after 27 seconds on the first trial. He tried for 63 seconds on the second trial, with random
**TABLE 15**  
**CASE 2: SUMMARY OF RESULTS**

Chronological age: 5 years, 8 months

<table>
<thead>
<tr>
<th>McCarthy Scales</th>
<th>Scaled Score</th>
<th>Percentile</th>
</tr>
</thead>
<tbody>
<tr>
<td>General Cognitive Index</td>
<td>50</td>
<td>1</td>
</tr>
<tr>
<td>Verbal Scale Index</td>
<td>28</td>
<td>1</td>
</tr>
<tr>
<td>Perceptual/Performance Index</td>
<td>22</td>
<td>1</td>
</tr>
<tr>
<td>Quantitative</td>
<td>22</td>
<td>1</td>
</tr>
<tr>
<td>Memory</td>
<td>22</td>
<td>1</td>
</tr>
<tr>
<td>Motor</td>
<td>26</td>
<td>1</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Preschool Language Scale</th>
<th>Age Equivalent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Verbal Ability</td>
<td>3-10</td>
</tr>
<tr>
<td>Auditory Comprehension</td>
<td>4-0</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Peabody Picture Vocabulary Test-Revised</th>
<th>Age Equivalent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Standard Score</td>
<td>67</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Developmental Test of Visual-Motor Integration</th>
<th>Age Equivalent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Standard Score</td>
<td>66</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Wallin Pegs-A</th>
<th>Time</th>
<th>Age Equivalent</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>17&quot;</td>
<td>3-0 to 3-5</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Wallin Pegs-B</th>
<th>Time</th>
<th>Age Equivalent</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>19&quot;</td>
<td>3-0 to 3-5</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Stutsman Buttoning Task</th>
<th>Button</th>
<th>Age Equivalent</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>1</td>
<td>Not completed</td>
</tr>
<tr>
<td></td>
<td>2 Buttons</td>
<td>Not completed</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Seguin Formboard</th>
<th>Time</th>
<th>Age Equivalent</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>1'47&quot;</td>
<td>3-0 to 3-5</td>
</tr>
</tbody>
</table>
attempts at buttoning, before he gave up. His performance was characterized by pushing on the button hole, lifting the top piece of fabric up to look, then pushing again. He did get part of the button through, but did not know how to complete the task further. The two button task was refused immediately.

**Case 3**

Case 3 is an active 5 year, 4 month old boy with a history of hydrocephalus, and an early diagnosis of cerebral palsy. He was observed to bunny-hop or scoot to get where he wants to go, pull to stand and walk if assisted.

His face is somewhat dysmorphic, with a prominent brow, a slightly recessed mandible, an upturned nasal tip and depressed bridge, wide set eyes with bilateral epicanthal folds. It has also been noted that he has low set ears. He has alternating esotropia.

His mother reports that her full term pregnancy was normal, although she had high blood pressure the last few weeks. Labor length was 11 hours, during which time the fetal heart rate dropped. At birth, his head circumference was noted to be larger than normal. Hydrocephalus was diagnosed, but shunting was not necessary. He was in a special care nursery for five weeks, ventilated and tube fed.
At 11 months a slight cervical and upper thoracic scoliosis was noted. A genetics evaluation indicated no chromosomal abnormalities. He has a history of asthma and ear infections. He underwent surgery for a bilateral inguinal hernia at the age of two, an umbilical hernia repair at the age of three, and had two surgeries to correct a club foot at the age of four.

Agenesis of the corpus callosum was confirmed by CT scan, which also indicates a slightly dilated posterior horn of the left lateral ventricle. At the age of 5 years, 1 month, Case 3 was admitted to the hospital with what appeared to be seizures. No diagnosis of seizure type is noted in the record.

Previous Assessments

An initial screening took place when Case 3 entered early intervention preschool at the age of 11 months. At that time he was rolling and attempting to sit. He was babbling in a consonant-vowel pattern and taking turns vocally. The ABSI indicated that he had moderate to severe delays in adaptive behavior. Concept skills were a strength, with relative weaknesses in physical development, communication, and play.

The EIDP indicated that he was functioning in the 3 to 5 month range in fine motor, and social emotional development. Cognition and gross motor abilities were solid
at the 3 to 5 month level and scattered to 6 to 8 months. Language was at the 6 to 8 month level.

At 23 months, the EIDP indicated that he was functioning at 6 to 8 months with some fine motor and gross motor skills at the 9 to 11 month level. Cognition and self care were at the 6 to 8 month level. Social-emotional development remained at the 3 to 5 month level, and language remained at the 6 to 8 month level. He tended not to imitate if an adult was looking. At that time, Case 3 was pulling to stand, and bunny-hopping to move around. He exhibited overall low tone. He tended to hit his chin as a self-stimulatory behavior.

Current occupational and physical therapy reports indicate that at the age of 5 years, 2 months, Case 3 is pulling to stand and cruising on furniture. He walks with assistance, and has a walker, but generally does not use it.

Tests Administered

Bayley Scales of Infant Development

The Bayley was administered due to Case 3's overall level of development. His performance indicates that his age equivalent on the mental scale was 9 months, with some skills in the 15 month range. The age equivalent on the motor scale was 9 to 11 months. His performance was significantly affected by his tendency not to look when manipulating objects.
On the mental scale, he used gestures to make his needs known and imitated a simple physical movement. He is reported to have one approximated sign. He vocalized three different syllables, and appeared to listen selectively to some words. He responded to simple verbal requests, and inhibited briefly to "no."

He held the crayon adaptively and made marks on the paper. He did not transfer objects hand to hand, and appeared to be tactually defensive with the left hand. He held a block in the left hand only briefly. His left arm tended to be held in high guard with the fingers partially fisted. He picked up a one inch cube directly, in spite of appearing to look in another direction. The pellet was regarded only briefly, and no attempt was made to retrieve it.

Objects that made noise were inspected with sustained interest. Most objects that had only tactual or simple visual properties were regarded briefly, then knocked away or thrown behind him. The book was regarded with interest, and he brought his face very close to look at it.

Socially, he was not spontaneously interactive. Interaction was brief, and he was easily distracted. He did repeat performances that were laughed at, and imitated a few sounds. The biggest positive response noted was to his reflection in the mirror. He laughed and made eye contact.
for the longest duration of the evaluation. He cuddled when
the examiner picked him up.

Motorically, he stood up by and cruised along
furniture. He used a bunny-hop from a W-sit position to
move around the room. He was observed to stand alone very
briefly. He combined toys briefly at midline, and played
pat-a-cake.

**Adaptive Behavior Scale for Infants and Early Childhood (ABSI)**

The ABSI was completed with his mother reporting and
indicates that he has moderate to severe deficits in
adaptive behavior. Relative strengths include self
direction and socialization. Independent functioning is a
relative weakness.

In the area of independent functioning, Case 3
exhibits active resistance to eating and drinking. He eats
ground up or mashed food due to his aversion to textures.
He does not feed himself. He would prefer to have a bottle,
and resists when a cup is used. He is not toilet trained,
and does not currently show awareness that his diaper is
wet or soiled.

In the area of physical development, he appears to
have adequate visual and auditory functioning. Case 3 has
good head control. His grasp is generally delayed, he holds
a crayon in a fist, and obtains small objects using a
palmar grasp. He is able to grasp a variety of objects, but often doesn't look while obtaining the object.

In the Language domain, Case 3 will point to some familiar objects in pictures if they are something he likes. He can follow simple one step directions. Expressively, he indicates needs using vocal sounds and gestures, although he occasionally will use a few signs or sign approximations.

Conceptually, Case 3 will watch and play with his hands, and seems to recognize himself in a mirror. In play, he engages in a variety of sensorimotor activities, including mouthing, banging, and participating in games such as peek-a-boo. He does engage in some exploratory play, including pushing toys, and dumping and filling containers. He enjoys physical play with adults.

In the self-direction domain, he attends to what is happening around him, tries to take part in other's activities, and will sometimes indicate a desire to do a certain activity. His usual attention span is reported to be one to five minutes. In the area of socialization, he will stay away from things he has been told not to touch and will inhibit to "no." He will comfort an unhappy person, recognizes friends and will indicate to an adult for help when needed.
TABLE 16
CASE 3: SUMMARY OF RESULTS

Chronological age: 5 years, 2 months

**Bayley Scales**

<table>
<thead>
<tr>
<th>Scale</th>
<th>Score</th>
<th>Age Equivalent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mental Development Index</td>
<td>N/A</td>
<td>9 to 15 months</td>
</tr>
<tr>
<td>Mental Scale Age Equivalent</td>
<td>N/A</td>
<td>9 to 15 months</td>
</tr>
<tr>
<td>Psychomotor Development Index</td>
<td>N/A</td>
<td>9 to 11 months</td>
</tr>
<tr>
<td>Psychomotor Age Equivalent</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Preschool Language Scale**

<table>
<thead>
<tr>
<th>Aspect</th>
<th>Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>Verbal Ability</td>
<td>1-1</td>
</tr>
<tr>
<td>Auditory Comprehension</td>
<td>1-1</td>
</tr>
</tbody>
</table>
He has some stereotyped behavior, including rocking his head back and forth, mouthing his hands, and hitting himself gently on his chin or other area of his face. He is reported to rarely sit still, and will excessively hang onto his mother for attention.

**Language Tests**

Preschool Language Scale. The PLS indicates that Case 3 has an age-equivalent of 13 months for verbal ability and auditory comprehension. He looked toward the door when asked where it was, and imitated some sounds. He did not attend to the pictures when presented, and does not request needs using words.

**Case 4**

Case 4 is a 2 year, 3 month old boy who was observed to move himself around using a baby walker, play with a car, and attempt to imitate a novel word. His face is generally symmetrical with deep set eyes, a prominent maxilla, and prominent ears.

Initial diagnosis was made in utero. An ultrasound at six months gestation was interpreted as hydrocephalus, due to the shape of the lateral ventricles. A chromosome analysis indicated 8P Trisomy, a syndrome which occasionally includes agenesis of the corpus callosum.
The pregnancy included early complications. Prior to eight weeks gestation, mother's hormone levels were not elevated as they should have been. The assumption was that the fetus had died. At eight weeks, the levels were checked again, the pregnancy was established and continued without further complication. Mother noted that she had some early spotting, but had a similar pattern during the previous pregnancy.

Case 4 was delivered at 41 weeks gestation after a four hour labor. During labor the fetal heart rate dropped. He spent 15 days in NICU, the first five on a ventilator, and was expected to die shortly after birth. The initial Apgar was 1. His birth weight was 6 pounds, 10 ounces.

He has had a series of significant health problems, the most severe of which was congestive heart failure, which was treated by medication throughout his first year and a half. No medication is currently prescribed. He has a coarctation of the aorta, which is being monitored, and may require later surgery. He also has a small ventricular septal defect.

Case 4 was tube fed through much of his early infancy, and later required oxygen during feedings. He has not been administered oxygen for over a year. He currently is fed baby food by mouth, and is highly resistive of texture. He was diagnosed as failure to thrive.
He had seizures shortly after birth, but has not had any since. He has had several ear infections, with tubes placed twice. He has had recurrent illnesses that include high fever, dehydration and vomiting. At 26 months, he had surgery to correct bilateral inguinal hernias. He also has had surgery to correct strabismus in his left eye.

His early milestones have been delayed. He rolled over at four months, but at 16 months had very little head control while held in a vertical position. He has never crawled. His means of mobility on the floor is rolling, scooting on his back, or pulling himself with his arms for short (less than one foot) distances. He is very mobile when placed in a baby walker. He does not currently use words to communicate, but word approximations are emerging.

**Tests Administered**

**Bayley Scales of Infant Development**

On the Bayley, Case 4 obtained Mental Developmental Index and Psychomotor Developmental Index scores of below 50. His Mental Scale age equivalent is 12 to 14 months, and his Motor Scale age equivalent is 9-11 months.

On the Mental Scale, activities that were completed unilaterally were with the left hand. He does bring objects to midline bilaterally, and plays pat-a-cake. He uncovered the cube by picking up the cup by its handle, but would put no cubes in the cup. He looked for the contents of the box,
but uncovered the box by knocking it over. He fingered the pegboard, and played with the peg, but did not place any. He has a tendency to look at an object, then look away before reaching for it. His accuracy is reasonably good.

He pushed the car, turned the pages of a book if given a slight start, and looked at the pictures in the book. He attended to the examiner's scribbling, held the crayon adaptively, but in spite of moves in that direction, never put the crayon to paper. He jabbers expressively, and says "Dada" and "Mama." On the investigator's initial visit, he imitated "stuck" with a close approximation and mother reported that he imitated it again a few days later.

On the Motor Scale, he sits alone steadily, but is unable to raise himself to that position. He stands up by furniture, and stands alone briefly. He was observed to stand for a longer period while in his walker, with little to no support. He will take a step when given support, but tends to lock his knees when standing.

He scooped the pellet, and used partial thumb opposition with the cube. He combines toys at midline. He throws a variety of objects, but does so randomly.
Adaptive Behavior Scale for Infants and Early Childhood (ABSI)

The ABSI was completed with mother as informant. Overall, his skills fall at the higher end of the moderate to severe range of adaptive behavior. Self-direction is a relative strength.

In the independent functioning domain, he eats baby food that contains little texture. He anticipates the spoon. His mother reports that he probably could hold his own bottle, but does not do so. He does not finger feed, and shows little interest in it. He is beginning to drink from a small cup. He is not toilet trained, and is only beginning to show awareness when his diaper is soiled.

In the area of physical development, he has visual and auditory awareness. It is not clear that he recognizes specific sounds, such as a telephone, but he does localize to sound. He has good head control, sits steadily, and needs some support to stand for more than a few seconds.

Case 4's receptive language skills include recognition of his own name and understanding simple gestures. Expressively, he indicates needs vocally, combines and imitates different sounds. He is just beginning to imitate words. He can indicate yes/no by shaking or nodding his head.

Conceptually, he watches his own hands and recognizes himself in a mirror. In the area of play, he stacks toys,
TABLE 17
CASE 4: SUMMARY OF RESULTS

Chronological age: 2 years, 3 months

Bayley Scales

<table>
<thead>
<tr>
<th>Scale</th>
<th>Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mental Development Index</td>
<td>50</td>
</tr>
<tr>
<td>Psychomotor Development Index</td>
<td>50</td>
</tr>
</tbody>
</table>

Preschool Language Scale

<table>
<thead>
<tr>
<th>Ability</th>
<th>Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>Verbal Ability</td>
<td></td>
</tr>
<tr>
<td>Age Equivalent</td>
<td>1-1</td>
</tr>
<tr>
<td>Auditory Comprehension</td>
<td></td>
</tr>
<tr>
<td>Age Equivalent</td>
<td>1-3</td>
</tr>
</tbody>
</table>
and plays with pull toys appropriately. He engages in a variety of sensorimotor play, and does some amassing. He enjoys exploratory and physical play with adults.

Case 4's strength is in self direction. He pays attention to his environment and tries to take part in the activities of others. He will concentrate on an activity that he likes for up to 15 minutes. In the area of socialization, he inhibits to "no," knows the names of friends and neighbors, and calls for help when he needs it.

Language Tests

Preschool Language Scale. The PLS was administered, and indicates an auditory comprehension age of 13 months and a verbal ability age of 15 months. Receptively, he understood a simple question. Expressively, he imitates a variety of sounds, and he uses a consistent label for his parents.

Case 5

Case 5 is a 2 year, 8 month old girl who is small for her age. She was observed to move smoothly, use five word sentences (example: "I'm going to sleep downstairs"), and ask questions. Her face is symmetrical, with wide set eyes, the appearance of epicanthal folds, and an open bite. Her brother has been identified as having a learning disability, and her father is reported to read slowly.
Prior to diagnosis, there had been no concerns about developmental delays. Her developmental milestones were within normal limits. They include rolling over at 5 months, crawling at 7 months and walking at 12 months. She said her first words at 12 months, and was toilet trained by 27 months.

Her medical history is generally unremarkable. Pregnancy was without complications, delivery was by scheduled repeat Caesarean section at 40 weeks. Birth weight was 7 lbs., 7 oz. She has had no serious illnesses, but has had reoccurring ear infections.

Case 5 was diagnosed with agenesis of the corpus callosum at the age of 2 years, 5 months. She had been dropped on her head by a relative. Two days after the accident, her mother noted swelling and tenderness, and had X-rays taken, with follow-up by an MRI. She sustained a non-depressed skull fracture in the left parietal area, with no loss of function reported.

Tests Administered

McCarthy Scales of Children's Abilities

Case 5 was somewhat uncooperative during administration of the McCarthy. Her attention span was short, she had difficulty separating from her mother, and she frequently refused to complete items. Her lack of cooperation was more a function of resisting demands than
inability to complete tasks, as she tended to complete
tasks she had refused at a later time. In spite of her
problems with cooperation, Case 5 obtained a GCI of 102.

As a whole, perceptual-performance skills were a
relative strength and verbal skills were relatively weak.
She had the most difficulty with verbal items that required
responses to the examiner's questions and did not include a
visual cue. The exception was items that required immediate
repetition of a cue; she could recall up to a three-item
string of information.

She could not recall any item on pictorial memory,
although she easily identified and labeled similar objects
with visual cues. She had trouble giving simple definitions
for objects or recalling items in a class. On all of these
items, she appeared to be trying hard to come up with an
answer for the examiner, but could not.

Case 5 seemed to enjoy fine motor tasks. She enjoyed
drawing, although she occasionally was resistive when asked
to draw something specific. On the tapping sequence, she
did not score beyond the first item because she tended to
hit the same key more than once before proceeding to the
next key.

She can copy a circle and vertical line. She drew a
face that was upside down, and was eventually obliterated
by scribbling. She can count with one-to-one correspondence
up to three, but is inconsistent to five. She has number concepts up to three. She discriminated simple sizes, shapes and colors.

She refused to imitate folding hands and twiddling thumbs. She did not sight through a tube. Handedness could not be established due to refusals, but it was observed that she tended to switch hands frequently. She used her left hand to draw shapes, and both hands to draw a person. She tended to use the hand closest to the site of the task, although she was observed to cross midline on several occasions.

When asked to catch the beanbag, she would stay two turns and try to leave. She can catch with two hands, and hit the target throwing right handed. She did walk a straight line and stood on her right foot. She initially refused to walk on tiptoe, and when she did, curled her toes under and walked on the tops of her toes. Her mother reported that she tends to do that when walking tiptoe. She refused to catch with one hand, throw with her left hand, stand on her left foot, and walk backward.

Adaptive Behavior Scale for Infants and Early Childhood (ABSI)

The subject's mother completed the ABSI. Her report indicates no significant to mild deficits in adaptive behavior. Strengths include independent functioning,
physical development, and self direction. Weaknesses include communication skills, concept skills, play, and personal responsibility and socialization.

She is reported to be independent in feeding, and toileting skills. She is able to dress and undress, but needs help with fasteners. She can go short distances from home without getting lost.

She has adequate sensory development and body control. She walks and runs well with only occasional falling. She jumps well, but is not able to hop on one foot.

In the area of language, Case 5 can follow two step directions. She uses sentences of five or more words, and can be understood by strangers most of the time. She does not use future tense, but does use plurals, possessives and pronouns. She asks a variety of questions, can carry on conversations, and can answer "where" questions. She appropriately answers yes/no questions. When asked, she can give her first name.

**Language Tests**

*Peabody Picture Vocabulary Test-Revised.* On the PPVT-R, she obtained a standard score of 90 and an age-equivalent score of 2 years, 3 months. She correctly identified pictures that represented a variety of objects. She had difficulty with items that showed secondary body parts.
Preschool Language Scale. On the PLS, Case 5 scored above her age level. Her auditory comprehension age equivalent was 3 years, 3 months, and her verbal ability age equivalent was 3 years, 4 months. The receptive skills she demonstrated included identifying action in pictures, distinguishing "on" and "under" and identifying objects by their use. She inconsistently discriminated size. When 10 or more visual cues appeared on one page, she appeared to get confused.

Expressively, she used plurals, repeated sentences of 5 words in length, and answered number questions to three. She completed one item requiring opposites. She was able to answer items asking what she would do to meet physical needs (i.e. hungry), but had difficulty answering a more abstract question ("Tell me about..").

Perceptual-Performance and Bimanual Tasks

Sequin Formboard. Case 5 completed the formboard in 2 minutes on the first trial. The second trial was completed in 1 minute, 59 seconds, which corresponds to an age equivalent of 30 to 35 months. She occasionally had trouble with similarly-shaped items and tended to try to force shapes that did not fit. She had consistent trouble with neglecting the shape at the lower right hand corner of the board.
Wallin Peg boards. Case 5 was very motivated to
complete the task. She was consistently a few seconds
slower than expected on Board A. She completed it in 26
seconds on the first trial which corresponds to an age
equivalent of 18 to 23 months. She was slightly faster than
expected for age on Board B. She took 24 seconds and 23
seconds, respectively on her two trials which corresponds
to 36 to 41 months.

Case 5 tended to start randomly, skip a few holes,
then fill in the remainder. Holes to the left of midline
tended to be filled with the left hand, and holes to the
right with the right hand, although crossing of midline was
observed.

Developmental Test of Visual-Motor Integration. On
the DVMI, Case 5 successfully copied a vertical line.
Instead of a horizontal line she drew a second vertical
line. She did not correct the error, in spite of prompts to
copy the cue. She drew another vertical line for the
circle, then corrected when prompted. Once the circle was
complete, she drew more circular lines around it. The
cross, left oblique line and square were all a series of
vertical lines. Standard scores are not available for
children of her age, but appropriate completion of two
forms results in an age-equivalent of 3 years, 2 months.
### TABLE 18
#### CASE 5: SUMMARY OF RESULTS

Chronological age: 2 years, 8 months

<table>
<thead>
<tr>
<th>McCarthy Scales</th>
<th>Scaled Score</th>
<th>Percentile</th>
</tr>
</thead>
<tbody>
<tr>
<td>General Cognitive Index:</td>
<td>102</td>
<td>55</td>
</tr>
<tr>
<td>Verbal Scale Index:</td>
<td>46</td>
<td>49</td>
</tr>
<tr>
<td>Perceptual/Performance Index</td>
<td>60</td>
<td>53</td>
</tr>
<tr>
<td>Quantitative</td>
<td>56</td>
<td>51</td>
</tr>
<tr>
<td>Memory</td>
<td>52</td>
<td>50</td>
</tr>
<tr>
<td>Motor</td>
<td>54</td>
<td>51</td>
</tr>
</tbody>
</table>

**Preschool Language Scale**

- **Verbal Ability**
  - Age Equivalent: 3-3

- **Auditory Comprehension**
  - Age Equivalent: 3-4

**Peabody Picture Vocabulary Test-Revised**

- Standard Score: 90
- Age Equivalent: 2-3

**Developmental Test of Visual-Motor Integration**

- Standard Score: N/A
- Age Equivalent: 2-10 to 3-2

<table>
<thead>
<tr>
<th>Task</th>
<th>Time</th>
<th>Age Equivalent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Wallin Pegs-A</td>
<td>26&quot;</td>
<td>2-0 to 2-5</td>
</tr>
<tr>
<td>Wallin Pegs-B</td>
<td>23&quot;</td>
<td>3-0 to 3-5</td>
</tr>
<tr>
<td>Stutsman Buttoning Task 1 Button</td>
<td>Not completed</td>
<td></td>
</tr>
<tr>
<td>Seguin Formboard</td>
<td>2'00&quot;</td>
<td>2-6 to 2-9</td>
</tr>
</tbody>
</table>
Stutsman buttoning task. On the buttoning task, Case 5 used both hands, but quickly became frustrated. She got the button halfway through the hole, then gave up. Her movements when trying to complete the task were somewhat random, and she did not seem to be able to come up with a strategy to finish buttoning when she got the button halfway through. She refused the other buttoning tasks.

Case 6

Case 6 is a 5 month old female who is alert and socially engaging. She is the only child in the family. The pregnancy was complicated by bleeding during the tenth week. There is speculation that a twin spontaneously aborted at that time. Labor and delivery were without complications, and her Apgar scores were 8 and 9.

She was diagnosed as having agenesis of the corpus callosum shortly after birth, but there had been concerns since a prenatal ultrasound indicated possible ventricular megaly. Diagnosis was made at two months of age by MR imaging to rule out Sturge-Weber syndrome. Other diagnoses include left frontal lobe atrophy and porencephaly, cortical blindness affecting vision in the left eye, and seizure disorder.
Originally, the seizures were diagnosed as myoclonic and treated with Phenobarbital and Depakene. During the course of the evaluation, Case 6 was hospitalized with an increase in seizures, which were then diagnosed as infantile spasms, and treated with ACTH.

In addition, she has a microphthalmic left eye with coloboma, a hemangianoma on her left foot and left nipple, and hypotonia. Her right side is generally weaker than her left.

**Tests Administered**

**Bayley Scales of Infant Development**

The Bayley was administered when Case 6 was five months old. It should be noted that this was during the course of ACTH, which may have adversely affected the results. Generally, she was observed to be less active, less socially interactive, less vocal, and more cranky than she had been prior to the ACTH. In addition, she had the characteristic edema and seemed to have lower tone.

In terms of her general behavior, Case 6 responded to social approach and showed continued interest in interaction. She tended to respond to interaction by brightening, smiling, laughing and vocalizing. She seemed to be more aware of strangers than previously observed and reacted with slight vigilance before warming up. Initially, she seemed contented and recovered quickly from fussing,
however, by the end of the evaluation, she became difficult to console.

Positioning was an issue throughout the evaluation. She tended to keep her head to the right of midline more frequently than previously observed. Her left hand is the one she uses, tending to neglect her right hand. Her left eye is not currently thought to have much vision. She was observed to play using her left hand in right hemispace while in a partial side-lying position on her right side.

On the Mental Scale, her Mental Developmental Index was 85. She watched the materials, and engaged in simple play with the rattle and the red ring. She tended not to reach for materials, except for the red block, and a familiar toy, which she activated repeatedly by touch. Blocks placed in both hands were retained briefly. She retained the block longer in her left hand than her right.

She oriented to sight and sound on both the left and right sides. She tracked objects in a variety of directions. When positioned adaptively, she is more likely to use eye-hand coordination when reaching.

She vocalized both spontaneously and when approached socially. More than two sounds were heard, and she vocalized pleasure (cooing) and displeasure (squeak-like sound).
On the Motor Scale, her Psychomotor Developmental Index was 85. She turned from her side to her back. When placed in prone, she elevated her head using her arms. She sits with slight support, with good head control. Some head lag was noted with change of positions. Her grasp varied between ulnar-palmar and partial thumb opposition.

Adaptive Behavior Scale for Infants and Early Childhood (ABSI)

The subject's mother served as informant on the ABSI when Case 6 was three months old. Generally, Case 6 is functioning with no significant deficits in adaptive behavior, with the exception of mild delays in physical development. Socialization is a relative weakness.

In the area of independent functioning, she is bottle fed, and cereal fed once per day. She opens her mouth in anticipation of feeding, and sucks and swallows effectively.

In the physical development domain, she shows visual awareness, looks at objects within her reach, inspects her feet, and looks at people across the room. She tracks objects in a variety of directions, and will look from object to object. She visually inspects objects and maintains eye contact with others. When lying on her stomach, she can lift her head. She tries to reach for objects, generally with her left hand due to the right sided weakness.
TABLE 19

CASE 6: SUMMARY OF RESULTS

<table>
<thead>
<tr>
<th>Chronological age: 0 years, 5 months</th>
<th>Bayley</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Mental Development Index</td>
</tr>
<tr>
<td></td>
<td>Psychomotor Development Index</td>
</tr>
</tbody>
</table>
In the area of communication, she seems to respond differently to different tones of voice. She babbles, and will imitate mouth movements. Conceptually, she watches and plays with her hands. In play, she visually follows people to where they disappear. She mouths and bats, attempts to pull another's clothing or hair and actively participates in peek-a-boo. She enjoys gentle roughhousing with adults.

In the area of self-direction, she attends to her environment, plays with toys suspended overhead for a long time, and has an attention span of one to five minutes. Socially, she appears to recognize people she sees frequently, and smiles and coos when interacted with by others.

**Subjects with Partial Agenesis**

**Case P7**

Case P7 is a 4 year, 11 month old girl who was observed to walk independently with poor balance, repeated a variety of phrases previously heard, and who explored her environment. She has a somewhat asymmetrical face, a prominent forehead, an open bite, and wide set eyes.

The pregnancy was complicated by edema. Case P7 was delivered by Caesarean section due to fetal cardio-pulmonary distress. According to the medical records, labor had not progressed after four days. Her
Apgar scores were 1 and 4. She required assisted ventilation for one day, and was hospitalized for 54 days.

A ventriculoperitoneal shunt was placed shortly after birth due to congenital hydrocephalus. A second shunt was placed when she was approximately one month of age, due to an arachnoid cyst. She has had two shunt dysfunctions, which have been corrected.

At the age of 4, she began having tonic-clonic seizures and is currently prescribed Phenobarbital. She has intermittent esotropia of the right eye. She has had repeated gastroenteritis and recurrent ear infections.

Agenesis of the corpus callosum was noted in her records as having been confirmed by CT scan and MR imaging. Reexamination of the films indicate that she has a partial corpus callosum. She is reported to have mild to moderate dilation of both lateral ventricles. There is evidence of polymicrogyria in the right parietal region and grey matter heterotopias. Developmental milestones have been delayed. Case P7 began walking at the age of 4 years, 4 months. Her mother recalled that she raised her head in prone at 13 months.

**Previous Assessments**

Case P7 has been enrolled in early intervention since the age of 5 months. An assessment was completed when Case P7 was 4 years, 0 months. The Vineland Adaptive Behavior
Scale indicated mild to moderate delays in adaptive behavior, with relative strengths in socialization skills (Standard score 55) and relative weaknesses in motor skills (Standard score 43). The Washington Preschool Profile was used to screen for developmental levels. At that time, her gross motor skills ranged from 0-12 months, fine motor skills were in the 12-24 month range, and self-help skills were in the 12-24 month range. Preacademic skills were determined to be at the 18 month level, receptive language skills ranged from 0-12 months, and expressive language skills were in the 6-8 month range.

Tests Administered

McCarthy Scales of Children's Abilities. The McCarthy was administered with limited success. Case P7 tended to avoid eye contact with the examiner and with most objects. She spoke in 4-5 word sentences, but her speech was rarely on topic, and tended to be repetition of commercials, songs, and other familiar phrases. The most cooperation was obtained when she was prompted to spell her name, as her mother reported she could do. When the first letter was prompted, she completed the rest of her name.

She mouthed the blocks and amassed them in one area. She became somewhat agitated during puzzle solving, and bit her hand in apparent frustration. She did not attend to items on pictorial memory. She used the stick as a tool on tapping sequence.
Case P7 did not repeat any words, and there was little motor imitation. She did take the tube, but instead of looking through it, she placed it at midline on her forehead. She did not appear to understand any of the instructions on the motor coordination tasks, but the line seemed to provide enough structure for her to take a few steps along it after a demonstration. She scribbled on the drawing tasks, and was slightly more likely to use her left hand than her right.

**Bayley Scales of Infant Development**

On the Bayley Mental Scale, Case P7 obtained an estimated age equivalent of 15 to 17 months. She has object permanence, and looks at pictures. She tends not to imitate on command, but will imitate an action at a later time. She holds a crayon adaptively, and scribbles in a linear manner. She uses words to make her needs known, and communicates in two or more word phrases. She will repeat a performance that is laughed at and will show an adult a favorite toy on request.

On the Motor Scale, Case P7 obtained an estimated age equivalent of 17 to 19 months. She uses a partial pincer to obtain small objects. She is reported to play pat-a-cake, and was observed to clap. She sits and stands independently. She walks alone, but is much steadier when

has hands held or is pushing something. She descends stairs
with assistance and ascends stairs independently using a railing.

**Adaptive Behavior Scale for Infants and Early Childhood (ABSI)**

The ABSI was completed with mother reporting. Her current functioning level is in the moderate to severe range, with independent functioning and physical development being relative weaknesses compared to the other scales.

In the independent functioning domain, she finger feeds, and can spoon feed if assisted hand over hand. Toilet training has been attempted with some success. She will occasionally use the toilet, but is generally in diapers. She can undress herself, with the exception of unbuttoning.

In the area of physical development, Case P7 has adequate visual and auditory awareness. She continues to have difficulty maintaining eye contact with others, but this has improved recently. She has no difficulty reaching or grasping. She walks as a primary means of locomotion, but her balance is poor and she falls frequently. She is able to walk upstairs alternating feet.

With communication, Case P7 recognizes her name and can follow a simple one-step direction. She indicates her needs and can communicate with others in 5 or more word
sentences. Caretakers understand her more than half the time. She uses words to indicate simple action and quantity and has recently begun to use possessives. She asks questions through intonations or by parroting questions she hears her mother say.

In the concept skills domain, Case P7 recognizes herself in the mirror, and will point to more than three body parts. She can match simple shapes and seems to know that money is to buy things.

In the area of play, she engages in a variety of exploratory play activities. Her mother reports no amassing play. She has been observed to use simple pretend play. She enjoys physical play with adults.

In the self-direction domain, Case P7 pays attention to her environment, tries to take part in the activities of others, and will indicate her desire to do certain activities. She will attend to a preferred activity for at least 10 minutes. In the personal responsibility domain, she will stay away from things she has been told to, but has developed a tendency to be sneaky, and will try to play with forbidden items. She will inhibit to "no". She will help with simple domestic tasks, and particularly enjoys the dustbuster. She knows the names of people she sees regularly, and will call to others if in trouble.
In the Maladaptive behavior domain, Case P7 will sometimes act rebellious, including throwing objects when told not to, and deliberately spilling food. She will chew on clothing, and tends to put everything in her mouth. She will bite her hand if frustrated. Currently, she is having a number of sleep problems including staying up most of the night, and tends not to sleep much more than two hours at a time.

Language Tests

Preschool Language Scale. The verbal ability portion of the PLS was completed, and corresponds with an age equivalent of 21 months. She was observed to echo or imitate some words. She used more than 10 words, and used them in at least 3 word combinations. She requested food and toys using one to two word phrases.

On the auditory comprehension scale, she was able to look toward a familiar object, but no other response to pictures or directions was obtained, corresponding to an age equivalent of 13 months.

Perceptual-Performance and Bimanual Tests

Wallin Pegboard. Administration of the pegs was attempted. Case P7 massed the pegs and tried to mouth them. No attempt to place them in the pegboard was observed.
**TABLE 20**

**CASE P7: SUMMARY OF RESULTS**

<table>
<thead>
<tr>
<th>McCarthy Scales</th>
<th>Scaled Score</th>
<th>Percentile</th>
</tr>
</thead>
<tbody>
<tr>
<td>General Cognitive Index:</td>
<td>N/A</td>
<td>N/A</td>
</tr>
</tbody>
</table>

**Bayley**
- Mental Development Index: N/A
  - Age Equivalent: 1-3
- Psychomotor Development Index: N/A
  - Age Equivalent: 1-5

**Preschool Language Scale**
- Verbal Ability
  - Age Equivalent: 1-9
- Auditory Comprehension
  - Age Equivalent: 1-1
Case P8

Case P8 is a 2 year, 4 month old girl who has had an extensive medical history. She has been observed to roll, scoot around the floor on her back and laugh at her reflection in the mirror. Her face is generally symmetrical with wide-set eyes and a relatively small jaw, her head is asymmetrical due to hydrocephalus. She lives with her parents and an older brother. Her father is a computer programmer and her mother had been employed as a waitress.

Her mother reports that there were no complications during the pregnancy. At 39 weeks gestation, an ultrasound was done due to concerns that there was a breech presentation. At that time, hydrocephalus was noted and a Caesarean section was performed the following day. At one day of age, a ventriculoperitoneal shunt was placed on the right side. She was hospitalized for three weeks.

Five days later, Case P8 was rehospitalized with a diagnosis of thrush. An NG tube was placed for supplemental feedings. She was hospitalized again at two months of age with a diagnosis of chalasia. At seven months of age, she underwent a coronal synostectomy. Surgery to correct esotropia of the right eye was performed at 14 months and further surgery was performed at 19 months. Background history indicates agenesis of the corpus callosum. Examination of the CT scan indicates the presence of a posterior portion of the corpus callosum.
Previous Assessments

Case P8 had an initial developmental assessment at 10 months, using the Early Intervention Developmental Profile. At that time, her gross motor skills were at the 2 month level. She is reported to have been able to lift her head for just a few seconds, and to dislike being placed in prone. Her range of motion was within normal limits, her head control poor in all positions, and her muscle tone was generally normal.

Her fine motor, cognitive and language skills were at the 3 to 5 month level. In fine motor, she presented with an ulnar palmar grasp emerging. She was able to track objects to the left, but was inconsistent to the right. Communication skills included repetition of syllables, differentiation of crying sounds and some imitation of mouth movements. She was beginning to turn to sound. She was described as very alert and social.

Adaptively, she was exhibiting mild delays overall on the ABSI, with significant weaknesses in physical development and communication, and strengths in concept skills and self-direction.

Another developmental assessment was conducted at 25 months using the Hawaii Early Learning Profile. At that time, she was passing cognitive items at the 9 to 11 month level. She was also performing some skills up to the 14
month level with help, including stacking rings and imitating gestures. Expressive language skills were at the 3 to 6 month level, with little change from the previous assessment. Her social-emotional skills were in the 12 to 15 month range, with skills including attempts at self direction, testing for parental reaction, and brief cooperative play. In her classroom, she activates cause-effect toys that have interesting visual and/or auditory spectacles, and is beginning to be introduced to a simple picture communication board.

**Tests Administered**

*Bayley Scales of Infant Development.* Very little cooperation was obtained from Case P8, and administration of the Bayley took place over several sessions. Overall, her Mental Developmental Index and Psychomotor Developmental Index are both below 50. Her performance indicates an age equivalent of 6-10 months on both scales. On the Mental Scale, items that involve social interaction or exploration of cause-effect materials are strengths. On the Motor Scale, her fine motor skills are at a higher level than gross motor.

Case P8 inconsistently used eye-hand coordination in reaching. Most materials were pitched after a brief inspection. She showed little interest in the ring, beyond an initial examination of it. She retained two blocks
briefly. She was not interested enough in the blocks to reach for the second or third when offered. She did attend briefly to the pictures in the book. She attended longest to interesting visual spectacles such as the mirror, and shiny objects. She fingered the pegboard shook the rattle, and rang the bell.

She has a playful response to the mirror, and enjoys brief social interaction. She will cooperate briefly with social games, can respond to simple requests, and vocalizes attitudes. Three different consonant-vowel syllables were observed. She rarely imitates, but enjoys being imitated.

On the Motor Scale, Case P8 can roll over, but prefers to be on her back. She continues to have a head lag in a variety of positions, but generally has good control in sitting. She appeared to have the control to sit briefly, but resisted by throwing herself to one side. She has a great deal of interest in moving, and can get from one end of the room to the other if she wants to. She does not bear weight on her legs, even when placed in a prone stander. She picked up the cube using partial thumb opposition, and was able to scoop the pellet, and an inferior pincer grasp appears to be emerging. She briefly combined objects at midline and can clap her hands together.
Adaptive Behavior Scale for Infants and Early Childhood (ABSI).

Case P8 is currently functioning with moderate to severe delays in adaptive behavior, according to her mother's report. Self direction is a relative strength, with independent functioning, physical development and communication being areas of weakness.

In the area of independent functioning, her eating skills are affected by her tube feedings, which comprise the majority of her daily intake. She does occasionally eat pureed food, but tends to thrust it out of her mouth with her tongue or gag when fed orally. She does not attempt to finger feed. Case P8 will open her mouth in anticipation of a bottle or spoon. Her suck and swallow are somewhat weak, and she can take thin liquids only. She is not toilet trained, but is reported to show some awareness when wet or soiled. She removes her own shoes and socks and assists with dressing by moving her arms and legs to help.

In the physical development domain, Case P8 exhibits visual and auditory awareness. She has been staring into space on an increasing basis. She is reported to have good head control, which has improved markedly in the past two months. She has motor control of reaching only when in certain positions, tends not to bring objects to her mouth and is reported to frequently miss objects that she is
reaching for. Case P8 need adaptive equipment for sitting and standing. She uses scooting on her back as her primary means of locomotion.

In the area of communication, Case P8 will respond differently to various tones of voice, and understands some simple gestures. She will indicate wants and needs using vocal sounds. She babbles a repeated sound and is just beginning to combine different sounds. When asked if she imitates, mother noted that people imitate Case P8, but she tends not to imitate them. Cognitively, she watches and plays with her hands, and appears to recognize herself in mirrors.

Her play skills include a variety of sensorimotor activities including batting, banging, and dropping objects. She actively participates in peek-a-boo. She is showing some interest in exploratory play with simple cause and effect toys that have interesting visual or auditory effects. She shows little interest in stacking objects, pull toys or ball play. With adults, she enjoys physical play.

In the area of self direction, she pays attention to what is happening around her, and will indicate a desire to watch television. She will attend to favorite videos for thirty minutes, but her attention span for most other activities is between one and five minutes. In the
<table>
<thead>
<tr>
<th>Table 21</th>
<th>CASE P8: SUMMARY OF RESULTS</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Chronological age:</strong> 2 years, 3 months</td>
<td></td>
</tr>
<tr>
<td><strong>Bayley</strong></td>
<td></td>
</tr>
<tr>
<td>Mental Development Index</td>
<td>50</td>
</tr>
<tr>
<td>Age Equivalent</td>
<td>6 to 10 months</td>
</tr>
<tr>
<td>Psychomotor Development</td>
<td>50</td>
</tr>
<tr>
<td>Age Equivalent</td>
<td>6 to 10 months</td>
</tr>
<tr>
<td><strong>Preschool Language Scale</strong></td>
<td></td>
</tr>
<tr>
<td>Verbal Ability</td>
<td></td>
</tr>
<tr>
<td>Age Equivalent</td>
<td>1-1</td>
</tr>
<tr>
<td>Auditory Comprehension</td>
<td></td>
</tr>
<tr>
<td>Age Equivalent</td>
<td>1-1</td>
</tr>
</tbody>
</table>
socialization domain, she will inhibit to "no", recognizes a variety of people, and will call out for help when she is in trouble.

She exhibits some problem behaviors, including refusing to eat, and temper tantrums. She has some self-abusive behavior, including chewing on her hand, slapping and banging her head, pulling her hair and scratching her face. The intensity and severity of these behaviors seem to increase with pain due to her gastrointestinal conditions. She tends to move constantly, frequently rocking her head side to side or scooting around the room. She frequently wakes up at night.

**Language Tests**

**Preschool Language Scale.** On the PLS, Case P8 passed the first item on the auditory comprehension which corresponds to an age equivalent of 13 months. She looked when asked where her teacher was. She also looked intently at pictures, but did not indicate that she knew what those pictures were. On the verbal ability scale, she imitated one sound, and appeared to imitate a second. Her age equivalent is less than 13 months.
CHAPTER VI
DISCUSSION

General Findings

It is not yet possible to attribute specific behavioral errors or developmental deficiencies directly to agenesis of the corpus callosum. It appears that there is a relationship between associated anomalies and functioning level in children with agenesis of the corpus callosum. Children with more serious developmental errors, such as chromosomal anomaly or hydrocephalus, tend to have lower functioning levels. Several children in this study had other factors, including anoxia, that could explain the presence of developmental delays. There tended to be a relationship between time spent in the hospital after birth and overall functioning level.

As with previous studies, there was little to no similarity in pregnancy complications that could be related to the agenesis, although three pregnancies had complications that could have been related. One mother had X-rays at approximately two weeks gestation and was on
medication for an infection throughout the pregnancy. A second child was thought to have spontaneously aborted just before 8 weeks gestation, due to low hormone levels. The levels later were more normal and the pregnancy continued with few complications. A third pregnancy had complications that were explained as the spontaneous abortion of a (possibly monozygotic) twin at 10 weeks.

Patterns of Developmental Delay

The initial research question asked whether specific patterns of delay could be identified. In spite of the initial heterogeneous appearance of the sample in this study, two general groups emerged, as hypothesized by Teeter and Hynd (1981).

The first group has cognitive levels ranging from the mild deficit range to the average range and adaptive levels from mild deficits to no significant deficits. They tended to have individual patterns of cognitive strengths and weaknesses. Adaptively, they tend to have weaknesses in concept and play skills, with strengths in independent functioning, self direction and physical development. These children, except for Case 5, have exhibited delays in language, motor and cognition from an early age. Case 5 has no related neurological damage identified, and had no birth complications.
The second group tended to be more medically involved, with more serious neurological sequelae. These children have cognitive levels below the mild deficit range and adaptive levels in the moderate to severe deficit range. Overall, their cognitive and motor skills tend to show similar degrees of delay. They tend to have considerable language delays, and they tend not to coordinate their vision and motor skills. Adaptively, their profiles were relatively flat with relative strengths in self direction and social skills and relative weaknesses in independent and physical functioning. Overall, their functioning levels were very similar to the two subjects who had partial callosal agenesis with other neurological anomalies.

There are also similarities across the subjects. They tend to have a short attention span and tend to be visually and auditorially distractible. They all had some degree of difficulty coordinating visual-motor skills. Some were subtle and others were readily apparent. They all appeared to have trouble encoding language, again, subtle in some cases and easily observed in others.

They were generally resistant to the structure imposed by standardized testing. This is likely to be in part, a function of their young age and developmental
level. However, there was also a strong qualitative sense that they preferred to be left to their own devices to figure the task out in their own way and in their own time.

The fourth research question asked: Are there differences in patterns of delays that could be a result of individual differences in addition to the extent of associated anomalies. The data appear to support this. Some children had relative strengths in visual-motor skills, while others had relative strengths in language. Adaptively, while the two general groups had similar patterns of strengths and weaknesses, there were individual variations within those patterns. While there were general similarities in factors such as distractibility and tendency to be self-directed in activities, each child had their own distinct temperament. At this point, the total number of young acallosal children studied is too small to avoid having some of the conclusions affected by individual differences.

It is suggested then, that as indicated by the literature, young children with agenesis of the corpus callosum tend to have general delays in areas such as language and fine motor skills. The degree of the delay is likely to be related to factors other than the agenesis.
Specific Developmental Delays

The second research question addressed more specific developmental delays. The first part of the question was: Will acallosal children display delays in visual-motor tasks and language. Visual-motor functioning and language will be considered separately below.

The second part of the question was: Will acallosal children show greater delays in visually aided bimanual tasks than bimanual tasks completed without visual input. This aspect of the question could not be addressed directly because the children refused to attempt the buttoning task while visually screened. However, visual attention to task is one aspect that separated the higher functioning group from the more developmentally delayed group.

Observations of the higher functioning group indicated that they tended to use vision in completing a variety of bimanual tasks. The subjects in the more delayed group, including those with partial agenesis, tended not to coordinate their visual and motor activities. When manipulating objects, they tended not to look at what their hands were doing. When reaching, they either glanced briefly then looked away or reached without looking directly at the object. Those that reached without looking appeared to be using peripheral vision.
The conclusion that can be drawn is the opposite of the question raised by Field and her colleagues (1978). Children with agenesis of the corpus callosum perform tasks more competently when coordinating visual input and bimanual tasks that acallosal children who do not. It may be that as the complexity of the task increases, visual input may be more confusing for acallosal children, but the data in this study cannot address that question. It may also be that other factors are involved, since children who tended not to coordinate their vision and motor skills were the ones with the most severe concurrent anomalies.

**Visual-motor functioning**

The visual-motor abilities of these subjects is similar to what has been found in other studies. They are able to complete a variety of tasks, but tend to do so more slowly than normal. The data here suggest that acallosal preschool children are disorganized in their approach to visual-motor tasks, although they are accurate in completing the tasks.

One indication of slowing of visual-motor processing was the beanbag catch on the McCarthy. Except for Case 5, the children tended not to move their bodies to accommodate variations in the throw, and tended not to move their hands or arms until hit by the beanbag.
The one task that none of the subjects in the higher functioning group was able to complete was buttoning. Again, their approach was disorganized. All the children gave up quickly and had strong reactions to attempting another trial. The original design of the study had a visually screened trial of the buttoning task, in an attempt to replicate the finding that Field and her colleagues (1978) observed. One of their subjects was more able to perform fine motor tasks when not watching her hands. All the subjects had a strong negative reaction to another trial of the buttoning that even when screened, no data could be obtained.

**Language skills**

Overall, the subjects in this study who were verbal tended to have difficulty with verbal memory, appeared to do best receptively if there were other contextual cues, and were most expressively communicative on self-initiated topics. The case with the most intact language abilities was Case 5, who had no other known complicating factors. Those children with total agenesis in the lower functioning group were nonverbal.

In general, scores on the PPVT-R were below age level scores obtained on the PLS Auditory Comprehension scale. This may indicate that the children needed more of a context to process the cues, or may have been distracted by
the pictures. The second hypothesis is less likely, as they were able to identify or label pictures in a group of more than four on the PLS and the McCarthy.

They tended to have difficulty with verbal memory items, and were more likely to respond to an item if there was a visual cue. They all had some degree of difficulty labeling, giving definitions and naming items in a class with auditory cues only. Case 5 in particular tended to look confused, and seemed to be trying hard to access an answer. The subjects did best if they initiated a verbal interaction.

Case 2 had a very difficult time understanding what was expected of him the first time a direction was given. This appears to account in part for the cognitive scores that were lower than expected given his adaptive behavior level, and some of his visual-motor scores. He tended to ask for information repeatedly until he was able to remember.

Overall, it appears that there may be a relationship between the agenesis of the corpus callosum and the ability to encode language and access verbal memory in young children. The literature suggests that these generalized delays become more specific as the child matures.

Age and developmental delay

There has been suggestion in the literature that as the child matures, compensation for deficits occurs, and
the developmental delays become less apparent. The third research question asked: Will younger children be more likely to exhibit identifiable delays than older children. Because this was not a longitudinal study, the question of the role maturity plays could not be addressed. The question was approached in two ways. First, older children were compared to younger children. Second, for those children who had previous developmental assessments, the archival data were compared to current data.

The older children in the current study tended to have lower measured intelligence levels than the younger children. The older children in the study also tended to have more severe concurrent anomalies and been more likely to have had more than one complication. Therefore, it appears that the relationship is more closely related to sample selection issues than whether compensation may or may not occur over time.

Intrasubject comparison over time confirms again that associated anomalies are likely to play a role. Case 1, who had birth complications has made slower than expected, but steady progress across developmental domains over time. She also had early standard scores that suggested functioning in the mild deficit range, while current scores suggest functioning in the low average range.

Case 2 had no specific complications noted. He has also made slower than expected progress over time, with
apparent periods of rapid progress in fine motor and language skills. Case 3, who has hydrocephalus and had birth complications, has made very little progress over time.

Anecdotal data on Case 5, who has no complicating factors, suggests that she has displayed no noticeable developmental delay in any area, and has continued to progress at an expected pace over time.

Implications of the Study

The most important implication derived from this study is that when isolated from other factors, agenesis of the corpus callosum may not have a significant impact on the functioning level of young children. It appears that other factors, including birth complications and associated anomalies have a greater impact on the child's developmental progress.

The questions this raises include: Does the agenesis of the corpus callosum cause the developmental delays observed in these subjects? In the process of fetal development, did the associated neurological anomalies play a role in the agenesis of the corpus callosum or just worsen its effects? Or, was there another factor during fetal development that resulted in both the agenesis and the related anomalies?

Further implications include:

1. The current study confirms that the absence of a
The corpus callosum is not by itself a predictor of overall functioning level. It appears that the presence and severity of concurrent neurological damage has the most influence on functioning level.

2. Prenatal developmental factors that result in agenesis of the corpus callosum are also likely to impact other developing systems in the fetus. The extent to which this occurs is likely to result in early viability issues in the infant, as well as associated neurological and physical malformations, which impact on the functioning level of the child.

3. This study also confirms that persons with callosal agenesis who function in the low average to average range have specific, sometimes subtle deficits that impact on language, motor and adaptive behavior development.

4. In young acallosal children, language delays appear to be global. That is, while it is not possible to measure specific deficits in very young children, the instruments used suggest generalized deficits in receptive and expressive language.

5. Similarly, in young acallosal children, visual-motor delays appear to be global, and in children with delays in the mild deficit range or above, the effect appears to be one of slowed processing time.
6. The presence of hydrocephalus in children with anomalies of the corpus callosum is a poor prognostic sign. Even children with only partial agenesis, who should have the structural means for interhemispheric processing, tend to have severe developmental delays. This suggests that accurate differential diagnosis of whether the child has hydrocephalus, ventricularmegaly or dilation of the ventricles due to the agenesis is very important.

**Implications for Education of Young Acallosal Children**

Implications for early intervention of children with agenesis of the corpus callosum include:

1. There is a need for regular evaluation of functioning levels across developmental domains, as rate of development cannot currently be predicted. The children in this study exhibited individual rates of progress.

2. Evaluation instruments need to be chosen with care to avoid making educational decisions based on a test that is heavily verbally or perceptually loaded. A heavily loaded or one-dimensional instrument would not be able to accurately capture the sometime subtle deficits that acallosal children may display.

3. Children who have made little progress over a period of a few years need to have a functional curriculum based on individual strengths and family needs. Emphasis on social interaction and the coordination of vision with object manipulation is important.
4. Directions for tasks need to be given in short phrases with as many additional cues as possible, including pictorial, gestural and signs. During task completion, allow the child to make his or her own accommodations in the process to facilitate the development of behavioral adaptations.

5. Allow the child to make choices in activities set up by an adult, to take advantage of strengths in self-direction while facilitating possible weaknesses in concept and/or independent functioning skills.

6. Provide short, frequent interaction with adults at the child's level. Start by imitating the child, and work toward the child imitating the adult, with the eventual goal of introducing cognitive-based toys and activities.

7. Particularly with infants and young toddlers, where the extent of visual functioning may not be known, provide high-contrast and brightly colored toys.

8. Provide an environment that has predictable routines, with consistent rewards and consequences.

Limitations of the Study

Several limitations of this study exist. First, interpretation of the results should be made with caution. Although efforts were made to obtain complete information about neurological anomalies, and results of CT scans and MR images were reviewed, it is likely that these instruments did not reveal the full extent of structural
damage. They cannot account for neuronal anomalies and heterotopias. It is likely that even those subjects with minimal associated anomalies do not have otherwise normal brains. This minimizes the generalizability of any trends noted in the results.

Second, while the current sample size is larger than in most previous research, the size of the sample is a clear limit to generalizability. Because of sample size, determination of statistically significant relationships was not possible, even through use of nonparametric measures.

Third, the range of ages and functioning levels, while important for other reasons, increased the amount of heterogeneity in the sample. This made analysis of specific strengths and deficits in cognition, language, and other areas more difficult.

Fourth, the age of the subjects limited both the instruments that could be used and the reliability of the results. Most neuropsychological, psychological, and linguistic instruments are normed on older children and adults. Further, measures of cognition and other functions are limited in their reliability when used with children under the age of six.

Fifth, two subjects were dropped when they could not be located. Both subjects were reported by referral sources to have been of low socioeconomic status, therefore
differential mortality may be have been a factor in this sample. 

Sixth, one factor that was not considered in this study was the effect of family ecology issues on the subjects. There may have been differences in family environments that also impacted on functioning level.

**Future Research**

This study indicates several further avenues of research. First, it will be important to follow up young children as they get older. This will provide a clearer picture of whether developmental patterns do exist in children with agenesis of the corpus callosum. It may also provide evidence of ways in which compensation might occur in addition to developmental mechanisms. The ability to predict later functioning levels can help with educational planning and give parents a more realistic understanding of what to expect with their child. Longitudinal research will be important to determine the effects of callosal agenesis on adult functioning levels.

Second, the present study indicates that research into specific areas of language functioning will need to continue. Further research with preschool children will help lay the foundation for current studies of language and reading that are being conducted with acallosal school age children and adults.
Third, the ability of a person with agenesis of the corpus callosum to integrate their vision and motor skills seems to be an important factor in later development. Studies of the development of visual-motor integration in acallosal children may provide clues to behavioral compensation and developmental processes.

Fourth, continued research into early developmental strengths and weaknesses may begin to indicate behavioral patterns that signal the potential of agenesis of the corpus callosum in undiagnosed children.

Fifth, the facial resemblance of the children bears further study. It may be possible to determine a pattern of craniofacial dysmorphias that will assist in determining the need for further diagnostic procedures.

Sixth, the presence of a normally functioning subject indicates the need for further study to determine the course of development for this type of subject.

Conclusions

When isolated as much as possible from other factors, agenesis of the corpus callosum produces mild behavioral effects in young children that are observed only by structured evaluation. These may include difficulties in processing receptive language or accessing memory when deprived of other cues, and difficulty in functionally crossing midline. Other deficits noted in subjects in this
study may be attributed to other factors, such as other neurological anomalies and anoxia.

Few concrete conclusions can be derived from this study. The data suggest that it is possible for very young children with callosal agenesis to display only subtle deficits, although this is not the case for the majority of subjects. There appears to be a relationship between concurrent neurological anomalies, early postnatal complications and functioning level. Lower functioning children were more likely to have more anomalies and complications. Higher functioning children generally had fewer complicating factors, and one subject who was functioning as expected for age had no other known complications. As with other studies with acallosal subjects functioning in the normal range, this rare occurrence raises more questions than can be answered with our current level of knowledge.
REFERENCES


