l. Cassandra Bac, hereby submit this original work as part of the requirements for the degree of Master of Science in Genetic Counseling.

It is entitled:
Investigation of Speech Delay in Individuals with 1p36 Deletion Syndrome

Student's name: Cassandra Bac

This work and its defense approved by:

Committee chair: Robert Hopkin, M.D.

Committee member: Ashley Brazil, M.S.

Committee member: John Lynn Jefferies, M.D., M.P.H.

Committee member: Ann W. Kummer, Ph.D.

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Investigation of Speech Delay in Individuals with 1p36 Deletion Syndrome

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by

Cassandra Bac
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Committee Chair:
Robert J. Hopkin, M.D.

Committee Members:
Ashley Brazil, M.S., L.G.C.
Ann Kummer, Ph.D., C.C.C.-S.L.P.
John L. Jefferies, M.D., M.P.H.
Abstract

Background: 1p36 deletion syndrome is a chromosomal terminal deletion syndrome, with a reported incidence ranging from 1/5,000-1/10,000. Current literature details a variety of severe medical and developmental problems associated with the deletion syndrome, many of which have been incompletely described. Specifically, speech impairment is considered to be almost universal among individuals with 1p36 deletion syndrome; however, there are no previous studies that clinically characterize the specific speech-language patterns and abilities within this population. The purpose of this study was to assess the communication abilities, as well as to describe the types of speech and language problems identified within this population. Methods: Speech-language evaluations designed to assess verbal impairment, intelligibility, pragmatics, feeding difficulty, hearing, receptive language, and expressive language were performed by a certified speech-language pathologist on twenty-eight individuals (aged 1-17) with 1p36 deletion syndrome. Results: 54% of participants (n=15) were verbal, specific verbal ability ranged from mildly to severely impaired. None of the participants were found to have age appropriate verbal ability. 5 participants were previously diagnosed with apraxia. Following study evaluation, 14 participants (50%) were identified as showing signs characteristic of apraxia; while only 5 (18%) demonstrated speech skills without concern for apraxia. Conclusions: Over half of our study population demonstrated verbal communication abilities, which directly contrasts with previous reports of absent expressive speech in the majority of people with 1p36 deletion syndrome. In addition, our results illustrate a high prevalence of apraxia characteristics in the speech of individuals with 1p36 deletion syndrome. This information has the potential to impact the type of treatment recommended for these individuals. Further investigation into the specific speech and language skills of individuals with this deletion syndrome is warranted with a larger patient population.
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Introduction

1p36 deletion syndrome has been recognized in recent literature as the most common chromosomal terminal deletion syndrome in humans, with incidence estimates ranging from 1/5,000 – 1/10,000 (Gajecka, Mackay, & Shaffer, 2007; Heilstedt, Ballif, Howard, Kashork, & Shaffer, 2003). In 2003, Heilstedt et al. reported that although deletion sizes varied in a sample of 61 individuals with 1p36 deletion syndrome, there was some clustering of breakpoints occurring at 4.0-4.5 Mb from the telomere; most de novo deletions were found to occur primarily on the maternal chromosomes (Heilstedt, Ballif, Howard, Lewis, et al., 2003). Several clinical features are associated with 1p36 deletion syndrome, most commonly including characteristic facial features (deep-set eyes, prominent chin, flat nasal bridge, large anterior fontanel, midface hypoplasia, long philtrum, and asymmetric ears), intellectual disability, hypotonia/motor delay, speech delay, vision and hearing problems, feeding issues, growth delay, behavior problems, seizures, and heart defects (Gajecka et al., 2007; Vieira et al., 2011).

While speech impairment is considered to be almost universal among people with 1p36 deletion syndrome, it has not been well characterized in the current literature. Several studies have reported a high incidence of speech problems in patients with 1p36 deletion syndrome, with delayed or absent speech present in roughly 98% of these individuals (Battaglia et al., 2008; UNIQUE). Most commonly, expressive language ranges from very poor to absent in most individuals with 1p36 deletion syndrome (Gajecka et al., 2007). In a study including 60 individuals with 1p36 deletion syndrome, Battaglia et al. (2008) reported absent expressive language in 75% of patients (n=45), usage of only a few isolated words in 17% of patients (n=10), and only 8% (n=5) of patients used two word phrases (Battaglia et al., 2008). Another study conducted by D’Angelo et al. (2010) reported that of the nine individuals identified with
1p36 deletion syndrome, all presented with delayed speech development. Specifically, expressive speech did not occur in these individuals prior to 4 years of age, and was predominately absent or limited to only a few isolated words (D'Angelo et al., 2010).

There have been multiple studies presenting data on the speaking capabilities of individuals with 1p36 deletion syndrome; however, there is currently no available literature that clinically characterizes their specific speech patterns, abilities, and difficulties. This study aims to fill this gap in the literature as well as to gain a better understanding of why these individuals display absent or delayed speech, which has the potential to impact the types of clinical speech therapy services these individuals are receiving.

**Methods**

**Participants and Procedures**

This was a cross-sectional descriptive study of a cohort of individuals with 1p36 deletion syndrome aiming to characterize the specific types of speech problems and the receptive and expressive language abilities within this population. Participants were recruited by e-mail through the 1p36 Deletion Support & Awareness Group, as well as posting electronically on the 1p36 Deletion Support & Awareness website, and locally through Cincinnati Children’s Hospital Medical Center (CCHMC). Criteria for inclusion in the study comprised of: 1) a diagnosis of 1p36 deletion syndrome, confirmed by genetic testing, 2) attendance at the 1p36 Deletion Support & Awareness Group national conference (July 2014, Cincinnati, OH), or 3) visiting the Speech Language Pathology Department at CCHMC for assessment during the time the study was conducted. Components of the research study included a pre-assessment survey (Appendix A) and evaluation by a certified speech-language pathologist. All pre-assessment surveys were completed by primary caregivers of the individuals with 1p36 deletion syndrome. Participants
were not excluded from the study if they were not able to complete all components of the assessment.

Twenty-eight individuals with 1p36 deletion syndrome (1 - 17 years old) participated in this study. Speech-language evaluations assessed the following domains: verbal impairment, intelligibility, pragmatics, feeding difficulty, hearing, receptive language severity, and expressive language severity. Assessment measures for each participant varied as determined necessary by the speech-language pathologist based on the child’s skill level, chronological age, and/or cognitive ability. All measures have been published with their reliability and validity accepted for scientific use.

Participants’ speech characteristics were further categorized by severity level (age appropriate, mild, moderate, moderately-severe, or severe) within each domain. Standardized scores for speech are unable to be determined until after age 3 years. For individuals under age 3 years, informal assessments of overall speech were used as the primary factor in determining the severity level within each domain.

All participants and/or their primary caregivers provided informed written consent for their participation in this study. This research was approved by the CCHMC Institutional Review Board (IRB #: 2014-1650).

Assessment Measures

*Clinical evaluation of language fundamentals-IV (CELF-IV)* (Semel, Wiig, Secord, Pearson Education Inc., & PsychCorp (Firm), 2003).

CELF-IV is a standardized assessment of receptive and expressive language skills in children and young adults aged 5-21 years. This assessment tool is made up of 18 subtests, each targeting a different speech and/or language ability. Eleven of these subtests were administered
during this study including: Word Classes, Concepts and Following Directions, Formulated Sentences, Recalling Sentences, Word Definitions, Understanding Spoken Paragraphs, Sentence Assembly, Semantic Relationships, Sentence Structure, Linguistic Concepts, and Word Structure. These subtests produce a CORE language, receptive language, and expressive language standard score. Completion time was approximately 30 minutes.

*Preschool Language Scale-5 (PLS-5)* (Zimmerman et al., 2011).

The PLS-5 is an interactive assessment of developmental language skills for infants and children birth through age 7 years. This test aims to identify receptive and expressive language skills in the areas of attention, gesture, play, vocal development, social communication, vocabulary, concepts, language structure, integrative language, and emergent literacy (PLS-5 Examiner’s Manual). The PLS-5 is norm-referenced and standardized on a representative national sample. Completion time ranged from 45-60 minutes.


The ROWPVT and EOWPVT are co-normed tests that are designed to help clinicians make accurate comparisons of a child’s (2-8 years) receptive and expressive vocabulary skills. This test yields percentile scores based on a representative sample of more than 2,000 individuals. Testing time was approximately 15-25 minutes.

*Oral and Written Language Scales-II (OWLS-II), Written Expression Scale (WES) and Reading Comprehension Scale (RCS)* (Carrow-Woolfolk & American Guidance Service., 1995).

Each of the scales in the OWLS-II assesses four language processes on four separate scales: lexical/semantics, syntax, pragmatics, and supralinguistics. Specifically, the WES measures the expressive aspects of written language and the RCS measures the receptive aspects
of written language. The OWLS-II provides a detailed and integrated assessment by looking at the same linguistic structures across four distinct language processes. This test is designed for children and young adults aged 5-21 years. Testing time ranged from 10-30 minutes per subtest.

*Goldman Fristoe Test of Articulation-2 (GFTA-2) (Goldman & Fristoe, 1969).*

The GFTA-2 assesses the articulation of individual phonemes (consonants) and phoneme clusters in all positions of single words in children and young adults aged 2-21 years. The GFTA-2 consists of 34 picture plates used to elicit 53 target words designed to assess consonant production in the initial, medial, and final positions of words. This is a published, norm-referenced test that yields standardized scores. Testing time was approximately 5 minutes.

*Receptive-Expressive Emergent Language Test-3 (REEL-3) (Bzoch, League, & Brown, 2003).*

The REEL-3 was designed to identify young children (birth-3 years) acquiring language at a significantly delayed pace, and to determine if there is a significant discrepancy between receptive and expressive processes of emergent language. This test has two core subtests, Receptive Language and Expressive Language and information is obtained based on a caregiver interview. The REEL-3 is norm referenced and yields standardized scores and percentile ranks. Testing time was approximately 20-30 minutes.

*Schedule for Oral Motor Assessment (SOMA) (Skuse, Wolke, & Reilly, 2000).*

The SOMA is an evaluation tool that describes feeding behaviors in infants by objectively assessing oral-motor function. This test has been shown to have a positive predictive validity greater than 90% and sensitivity greater than 85% for the detection of infants with clinically significant oral-motor dysfunction (Skuse et al., 2000). Testing time was approximately 20 minutes.
**Pure Tone Screening and Impedance**

Hearing was assessed using a pure-tone audiometric screening. If a full audiological evaluation was completed on the participant within the past 12 months and hearing was found to be within normal limits, those results were substituted for a screening. Testing time was approximately 10 minutes.

**Tympanometry (Onusko, 2004).**

This test measures the ear’s response to both sound and pressure. This test involves placing a probe-type device into the ear canal, which changes the air pressure in the ear, making the eardrum move back and forth. Results from this test were measured on a tympanogram using decapascals (daPa). Testing time was approximately 5 minutes.

**Data Analysis**

Recruitment of participants and distribution of pre-assessment surveys took place between May 2014 and August 2014. Speech-language study evaluations were conducted between July 2014 and August 2014. Summary statistics were calculated using JMP10 Statistical Discovery Software to characterize the speech-language impairment within this population. Frequencies and proportions were reported for each categorical variable. Categorical variables were compared using a Fisher’s exact test. A statistical threshold of $p < 0.05$ for significance was used to complete data analysis.

**Results**

**Study Population**

A total of 28 individuals with 1p36 deletion syndrome were included in this study. Twenty two individuals were female (79%) and 6 were male (21%). For females, ages ranged from 1-17 years old, with an average age of 9.7 years. For males, ages ranged from 4-8 years old,
with an average age of 6.3 years. Twenty two individuals (79%) reported a previous diagnosis, other than 1p36 deletion syndrome. Categories were not mutually exclusive and some individuals reported more than one diagnosis. This information is summarized in Table 1. Twenty seven of the total 28 participants (96%) reported residency in the United States and one individual reported residency in Mexico. Information on deletion size was provided by 8 participants; summary information is found in Table 2.

**Speech and Communication**

Table 3 shows the results of the clinical speech-language evaluations. None of the participants were found to have age-appropriate verbal ability and none had age-appropriate receptive or expressive language abilities. Approximately 54% of participants (15 participants; 13 females and 2 males) were verbal, although specific verbal ability ranged from mildly to severely impaired. Twenty one participants (75%; 17 females and 4 males) had severe verbal impairment, 3 participants (11%; 1 female and 2 males) had moderately-severe impairment, 2 females (7%) had moderate impairment, and 2 females (7%) had mild impairment. The receptive and expressive abilities of participants are summarized in Table 4.

Of the total 28 participants, 5 participants (18%; 4 females and 1 male) were previously diagnosed with apraxia. Following study evaluation, 14 individuals (50%; 11 females and 3 males) were identified as showing signs characteristic of apraxia; while only 5 individuals demonstrated speech skills without concern for apraxia. The remaining 9 individuals (32%) were completely non-verbal.

Two out of the total 28 participants (7%; 2 females and 0 males) were found to have intelligible verbal language, defined as being comprehensible or able to be understood by the individual’s parents and speech-language pathologist. Intelligibility was unable to be determined
in 9 participants (32%; 6 females and 3 males) due to the child’s skill level, chronological age, and/or cognitive ability. Approximately 61% of participants (17 participants; 12 females and 5 males) reported using some form of alternative communication, including gestures, American Sign Language (ASL), Picture Exchange Communication System (PECS), an iPad, and the Dynavox and Tobii augmentative communication devices (Table 5). Categories were not mutually exclusive. Of the 17 participants who reported utilization of an alternative type of communication, 8 (59%) were non-verbal and used this as their only mode of expressive language. The remaining 9 participants also used verbal speech.

With regards to feeding, the parents of eleven participants (39%) reported feeding concerns, and 5 of these individuals had significant feeding concerns which required a gastrostomy or nasogastric tube. In the category of hearing, 9 participants (32%) had hearing concerns as determined by the audiologist, while the remaining 19 participants (68%) had hearing within normal limits. In addition, 9 participants (32%) demonstrated age-appropriate pragmatic skills, while the remaining 19 participants (68%) had pragmatic language skills that ranged from mildly to severely impaired (Table 3).

**Discussion**

Speech impairment is considered to be an almost universal phenotypic characteristic of 1p36 deletion syndrome. Current literature suggests that the majority of individuals present with absent expressive language, while some may only be able to speak and understand a few isolated words. Many of these previous studies have also presented information regarding the negative impact that this impairment has on the individual’s ability to have meaningful interactions with their family members and peers (Battaglia et al., 2008; D'Angelo et al., 2010; Gajecka et al., 2007; UNIQUE). While general speech delay/impairment was found to be universal among our
study population, detailed findings from our study challenge previous assumptions regarding the specific verbal and communication abilities within this population.

It has been previously reported that expressive language is more significantly impaired when compared to receptive language, with expressive language typically very poor or absent in most individuals with 1p36 deletion syndrome (Gajecka et al., 2007). While there have been reports of individuals with small deleted regions and more complex speech abilities, these individuals are considered to be the minority and do not represent the more commonly reported 1p36 deletion syndrome phenotype (UNIQUE; Wu et al., 1999). Specifically, Battaglia et al. (2008) reported absent expressive language in 75% of participants, and language limited to a few isolated words or at the level of two-word phrases in the remaining 25% (Battaglia et al., 2008). Over half of our study population demonstrated verbal communication abilities, which directly contrasts with previous findings of absent expressive speech in the majority of people with 1p36 deletion syndrome. Additionally, of those who were able to speak, 4 participants (14%) in our study showed only mild to moderate verbal impairment. Results from our study did not show a statistically significant association between receptive and expressive language skills, however, overall receptive language skills within our sample population were found to be better than expressive language skills.

There is general agreement in previous literature regarding the prevalence of speech delay/impairment within this population; however, there has been no detailed study that clinically characterizes the specific speech problems observed. Our results illustrate a high prevalence of apraxia characteristics in the speech of patients with 1p36 deletion syndrome. While only five participants in our study reported a previous clinical diagnosis of apraxia, half of our cohort was found to show signs and symptoms characteristic of apraxia. This information has
the potential to impact the type of speech-language therapies recommended for these individuals, which may allow for earlier implementation of appropriate and effective interventions. Results reported within this study highlight that health care providers, most importantly speech-language pathologists, must be aware of the possibility of apraxia of speech within this population, so that individuals are appropriately evaluated and treated.

In addition, our study revealed that verbal communication is not the only method of communication that is utilized by this population. Over 60% of participants reported using an alternative method of communication, other than verbal language. Of those, 59% used this as their sole form of expressive language. This finding demonstrates that despite the speech and language challenges, many individuals with 1p36 deletion syndrome are able to engage in communication, which can lead to development of meaningful relationships with family members and peers. Our results clearly demonstrate that individuals with 1p36 deletion syndrome are able to communicate in a variety of ways, and appropriate treatments and intervention strategies should be provided in order to maximize these communication skills.

It has also been reported that the majority of individuals with this deletion syndrome present with hearing deficits. Specifically, Gajecka et al. (2007) reported 77% of their cohort demonstrated hearing loss, either conductive, sensorineural, or both (Gajecka et al., 2007). The results from our study greatly contrast these previous findings in the literature. Following auditory evaluation, 69% of participants in our study were found to have hearing within the normal limits. Further investigation is warranted to examine these discrepancies. Feeding concerns in our cohort were consistent with previous reports.
**Limitations and Conclusions**

There are several limitations to this study. First, the complete documentation of genetic testing was accessible for only 8 study participants. Therefore, we were unable to make any conclusions with regard to genotype-phenotype correlations within our sample population. Second, due to age limitations for children under the age of 3 (7% of participants), standardized testing scores were unable to be formally calculated. Third, this study represents a small sample size, which should be considered when generalizing results to the larger 1p36 deletion syndrome population. Based on the results of this study, further investigation into the specific speech and language skills of individuals with this deletion syndrome is warranted with a larger patient population. In addition, it may be important to look at the neurological status of these patients in future research studies due to the known association of 1p36 deletion syndrome with a variety of clinical features including speech impairment, intellectual disability, hypotonia, seizures, and motor delay.

While previous studies describing the 1p36 deletion syndrome phenotype have broadly examined the speaking capabilities within this population, this study is the first to focus specifically on the speech problems observed within this population. Our findings suggest that apraxia of speech is a possible factor causing speech impairment and delay in individuals with 1p36 deletion syndrome. This awareness is extremely important for care providers, particularly speech-language pathologists, and may have implications for the type of treatment and intervention strategies that are recommended for the individual. Furthermore, our findings suggest that individuals with 1p36 deletion syndrome are, in fact, able to communicate in a variety of different ways, and implementation of appropriate treatment and intervention strategies has the potential to maximize these communication skills.
### Table 1. Reported Diagnoses (other than 1p36 Deletion Syndrome)

<table>
<thead>
<tr>
<th>Category</th>
<th>Specific Diagnosis</th>
<th>Males n=6 [%]</th>
<th>Females n=22 [%]</th>
<th>Total n=28 [%]</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Epilepsy</td>
<td>5 [83]</td>
<td>5 [23]</td>
<td>10 [36]</td>
</tr>
<tr>
<td></td>
<td>Hypotonia</td>
<td>2 [33]</td>
<td>8 [36]</td>
<td>10 [36]</td>
</tr>
<tr>
<td>Vision</td>
<td>Strabismus</td>
<td>0 [0]</td>
<td>4 [18]</td>
<td>4 [14]</td>
</tr>
<tr>
<td></td>
<td>Pulmonary Stenosis</td>
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<td>0 [0]</td>
<td>1 [4]</td>
</tr>
<tr>
<td>Gastrointestinal</td>
<td>Dysphagia</td>
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<td>0 [0]</td>
<td>1 [4]</td>
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<td>GERD</td>
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<td>1 [4]</td>
</tr>
<tr>
<td></td>
<td>Pierre-Robin Sequence</td>
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<td>2 [9]</td>
<td>2 [7]</td>
</tr>
<tr>
<td></td>
<td>Glossoptosis</td>
<td>1 [17]</td>
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<td>1 [4]</td>
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</table>
### Table 2. Chromosomal Deletion Information

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<tr>
<th>Participant ID</th>
<th>Cytogenetic Report Results</th>
<th>Method</th>
</tr>
</thead>
<tbody>
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<td>1p36003</td>
<td>del 1p36.33-1p36.32</td>
<td>SNP Microarray</td>
</tr>
<tr>
<td>1p36005</td>
<td>del 1p36.33-1p36.32</td>
<td>FISH</td>
</tr>
<tr>
<td>1p36007</td>
<td>del 1p36.33-1p36.32</td>
<td>SNP Microarray</td>
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<tr>
<td>1p36012</td>
<td>del 1p36.11-1p35.3</td>
<td>SNP Microarray</td>
</tr>
<tr>
<td>1p36014</td>
<td>del 1p36.33-1p26.31</td>
<td>Chromosomal Microarray</td>
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<tr>
<td>1p36020</td>
<td>del 1p36.33-1p36.23</td>
<td>Array CGH</td>
</tr>
<tr>
<td>1p36023</td>
<td>del 1p36.33-1p36.32</td>
<td>SNP Microarray</td>
</tr>
<tr>
<td>1p36025</td>
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<td>Array CGH</td>
</tr>
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</table>

### Table 3. Speech Language Evaluation Results

<table>
<thead>
<tr>
<th>Speech-Language Skill</th>
<th>Males n= 6 [%]</th>
<th>Females n= 22 [%]</th>
<th>Total n= 28 [%]</th>
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</thead>
<tbody>
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<td>Verbal</td>
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<td>13 [59]</td>
<td>15 [54]</td>
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<tr>
<td>No</td>
<td>4 [67]</td>
<td>9 [41]</td>
<td>13 [46]</td>
</tr>
<tr>
<td>Intelligible</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>0 [0]</td>
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<td>2 [7]</td>
</tr>
<tr>
<td>No</td>
<td>3 [50]</td>
<td>14 [64]</td>
<td>17 [61]</td>
</tr>
<tr>
<td>Receptive Language Age Appropriate</td>
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<td></td>
<td></td>
</tr>
<tr>
<td>Yes</td>
<td>0 [0]</td>
<td>0 [0]</td>
<td>0 [0]</td>
</tr>
<tr>
<td>No</td>
<td>6 [100]</td>
<td>22 [100]</td>
<td>28 [100]</td>
</tr>
<tr>
<td>Expressive Language Age Appropriate</td>
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</tr>
<tr>
<td>Yes</td>
<td>0 [0]</td>
<td>0 [0]</td>
<td>0 [0]</td>
</tr>
<tr>
<td>No</td>
<td>6 [100]</td>
<td>22 [100]</td>
<td>28 [100]</td>
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<tr>
<td>Feeding Concerns</td>
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<td>Hearing Concerns</td>
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<td>Yes</td>
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<td>9 [32]</td>
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<td>19 [69]</td>
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<td></td>
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<td>Yes</td>
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<td>9 [32]</td>
</tr>
<tr>
<td>No</td>
<td>3 [50]</td>
<td>16 [73]</td>
<td>19 [69]</td>
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Table 4. Receptive and Expressive Language Severity

<table>
<thead>
<tr>
<th>Time</th>
<th>Males n= 6 [%]</th>
<th>Females n= 22 [%]</th>
<th>Total n=28 [%]</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Receptive Language Severity</strong></td>
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<td></td>
<td></td>
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<tr>
<td>Age appropriate</td>
<td>0 [0]</td>
<td>0 [0]</td>
<td>0 [0]</td>
</tr>
<tr>
<td>Mild impairment</td>
<td>0 [0]</td>
<td>5 [23]</td>
<td>5 [18]</td>
</tr>
<tr>
<td>Moderately-severe impairment</td>
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<td>7 [31]</td>
<td>8 [29]</td>
</tr>
<tr>
<td>Severe impairment</td>
<td>2 [33]</td>
<td>5 [23]</td>
<td>7 [25]</td>
</tr>
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</table>

<table>
<thead>
<tr>
<th>Time</th>
<th>Males n= 6 [%]</th>
<th>Females n= 22 [%]</th>
<th>Total n=28 [%]</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Expressive Language Severity</strong></td>
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<tr>
<td>Age appropriate</td>
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<td>0 [0]</td>
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<tr>
<td>Mild impairment</td>
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<td>2 [7]</td>
</tr>
<tr>
<td>Moderate impairment</td>
<td>0 [0]</td>
<td>2 [9]</td>
<td>2 [7]</td>
</tr>
<tr>
<td>Severe impairment</td>
<td>4 [67]</td>
<td>17 [77]</td>
<td>21 [75]</td>
</tr>
</tbody>
</table>

Table 5. Alternative Forms of Communication Utilized by Participants

<table>
<thead>
<tr>
<th>Expressive Language Mode*</th>
<th>Males n= 6 [%]</th>
<th>Females n=22 [%]</th>
<th>Total n=28 [%]</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gestures</td>
<td>3 [50]</td>
<td>5 [23]</td>
<td>8 [29]</td>
</tr>
<tr>
<td>PECS a</td>
<td>2 [33]</td>
<td>3 [14]</td>
<td>5 [18]</td>
</tr>
<tr>
<td>ASL b</td>
<td>3 [50]</td>
<td>5 [23]</td>
<td>8 [29]</td>
</tr>
<tr>
<td>iPad</td>
<td>0 [0]</td>
<td>3 [14]</td>
<td>3 [11]</td>
</tr>
</tbody>
</table>

*Categories were not mutually exclusive.

a Picture Exchange Communication System
b American Sign Language

14
References


UNIQUE. 1p36 Deletion Syndrome (2011 ed.).


Appendix A: Pre-Assessment Questionnaire

Please answer each question as thoroughly as possible, even if your child is not receiving any of the therapies in question. If a question is not applicable to your child, please answer the questions with N/A. Sign and date at the bottom of page 9 and return pages 1-9 to our office.

Child Name:__________________________________________________________________________________

Street:_______________________________ City:_________________ State:_________ Zip:___________
Phones: Home:____________________ Cell:____________________ Work:____________________
E-Mail Address:___________________________________________________________________________

Please complete this section only if the child is a □ minor or an □ adult under legal guardianship:

Parent/Guardian:__________________________________________________________________________

Street:_______________________________ City:_________________ State:_________ Zip:___________
Phones: Home:____________________ Cell:____________________ Work:____________________
E-Mail Address:___________________________________________________________________________

Family History
Mother ___________________________________________________________ Age ________ Occupation ________________

History of Speech, Language, or Hearing Problems Yes ________ No ________
If “yes” please explain. _______________________________________________________________________

Father ___________________________________________________________ Age ________ Occupation ________________

History of Speech, Language, or Hearing Problems Yes ________ No ________
If “yes” please explain. _______________________________________________________________________

List names and ages of brothers and sisters:

Is there a family history of any of the following?

<table>
<thead>
<tr>
<th>Family Member</th>
<th>Family Member</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hearing loss</td>
<td>Cleft palate</td>
</tr>
<tr>
<td>Speech problems</td>
<td>Seizure disorder</td>
</tr>
<tr>
<td>Prematurity</td>
<td>Mental illness</td>
</tr>
<tr>
<td>Blindness</td>
<td>Alcoholism</td>
</tr>
<tr>
<td>Educational</td>
<td>Low birth weight</td>
</tr>
<tr>
<td>Drug use</td>
<td>Delayed motor development</td>
</tr>
<tr>
<td>Malformation of the neck or ears</td>
<td>Other</td>
</tr>
</tbody>
</table>

Child living with both parents _________ If no, whom does child live with ________________________
Have there been any of the following major changes in the family during the last year?

- change of address
- accident or illness
- divorce/marriage
- parent separation
- death of a family member
- birth/ adoption

Does anyone in the home smoke? Yes _______ No _______

Birth History

Mother’s health during pregnancy (note special conditions such as mumps, German measles, x-rays, serious accidents, etc.)

_________________________________________________________________________________________

Anything unusual about the condition of the infant at birth: Blue Baby _______ Lack of Oxygen _______
Convulsions _______ RH Problem _______ Breathing Difficulties _______ Head Injuries _______
Other (describe) __________________________________________

Length of pregnancy ____________________________  Birth weight of infant _____________________

Did child pass the newborn hearing screening?

Medical History

Please list all diagnoses your child has received:

________________________________________________________________________________________

Other Medical Information

Surgeries: __________________________________________

Childhood illnesses and injuries. Check each item that applies:

<table>
<thead>
<tr>
<th>Pneumonia</th>
<th>Convulsions</th>
<th>Concussions</th>
<th>Tonsillitis</th>
<th>Ear infections</th>
<th>Measles</th>
<th>Bronchitis</th>
<th>Enlarged adenoids</th>
<th>Other ear problems</th>
<th>Chickenpox</th>
<th>Asthma</th>
</tr>
</thead>
<tbody>
<tr>
<td>[ ]</td>
<td>[ ]</td>
<td>[ ]</td>
<td>[ ]</td>
<td>[ ]</td>
<td>[ ]</td>
<td>[ ]</td>
<td>[ ]</td>
<td>[ ]</td>
<td>[ ]</td>
<td>[ ]</td>
</tr>
</tbody>
</table>

Allergies (describe)  

Serious Injuries (describe)  

Other (describe)  

If your child has had fevers, how long do they last?  

Check any of the following drugs that your child has taken: Quinine _______ Streptomycin _______
Nicotine _______ Frequent aspirin _______ Neomycin _______

Child’s present health  

Has the child had an eye examination? _______ When? _______ By whom? _______

Name any medicines the child is currently taking:  

________________________________________________________________________________________
Primary Care Pediatrician

Name: ____________________________________________

Street: __________________________________ City: ___________ State: ________ Zip: ________

Phone: ___________ Fax: ___________ E-Mail: ____________________________________________

Developmental History

a. Hearing

Describe any hearing difficulties: _______________________________________________________

Has the child had hearing tests? ____________ When? ____________ By whom? ______________

What were the results of the hearing test? _____________________________________________

Does the child have a hearing aid? ___________ Does s/he use it? ____________

b. Feeding

Has your child had any feeding difficulties? Check each item that applies.

☐ Sucking or nursing

☐ Excessive length of time to drink bottle

☐ Regurgitation of liquids or solids through the nose

☐ Difficulty chewing or swallowing meats

☐ Choking and/or gagging

Does your child choke while eating? ☐ Yes ☐ No

If “yes,” on what foods? ___________________________________________________________

Is your child a picky eater? ☐ Yes ☐ No

If “yes,” what foods does s/he prefer? ________________________________________________

Describe any feeding problems your baby experienced during the first three months of life.

__________________________________________________________________________________

__________________________________________________________________________________

__________________________________________________________________________________

Does your child drool more than other children his/her age? ☐ Yes ☐ No

Did your child have difficulty gaining weight as an infant? ☐ Yes ☐ No
Describe any abnormalities of response to light, sound and movement ________________________________

______________________________________________________________________________________

At approximately what age did your child achieve the following motor milestones?

Head support _______ Reach & grasp _______ Sitting alone _______
Crawling _______ Standing alone _______ Walking alone _______
Climbing stairs _______ Finger food _______ Eat with a spoon _______
Potty trained _______ Undresses self _______

Child’s coordination: (Please check one)  ☐ Normal?  ☐ Fair?  ☐ Poor?

Right or left handed? ________________________________________________________________

At what age did handedness develop? __________________________________________________

Did anyone try to influence handedness? (describe) _______________________________________

Any abnormalities in early physical development? ________________________________________

**c. Play Behaviors**

Do you have any concerns with play behaviors?  ☐ Yes  ☐ No

Which of the following describes the type of play your child likes to engage in most often? (Please check all that apply)

☐ putting toys in mouth  ☐ banging toys together  ☐ throwing toys
☐ shaking toys  ☐ pushing/pulling toys  ☐ make believe play
☐ appropriate use of objects  ☐ uses one object for another  ☐ looking at books
☐ acting out familiar routines  ☐ role-playing  ☐ games with rules
☐  ☐ rough and tumble play

What is the average length of time your child can stay playing at one activity?  _______________________

**d. Speech History**

What languages are spoken at home? _______________________________________________________

Which are spoken by the child? __________________________________________________________

Which are understood by the child? ______________________________________________________

Indicate when your child first demonstrated the following:

<table>
<thead>
<tr>
<th>Age</th>
<th>Behavior</th>
<th>Age</th>
<th>Behavior</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>cooing, pleasure sounds</td>
<td></td>
<td>single words</td>
</tr>
<tr>
<td></td>
<td>babbling (ba-ba, da-da, etc.)</td>
<td></td>
<td>phrases (go bye-bye, more juice)</td>
</tr>
<tr>
<td></td>
<td>jargon</td>
<td></td>
<td>short sentences</td>
</tr>
</tbody>
</table>
What is the primary method(s) your child uses for letting you know what s/he wants? (Please check all that apply.)

- looking at objects
- pointing at objects
- gestures
- crying
- vocalizing/grunting
- physical manipulation
- single words
- 2-3 word combinations
- sentences

Which of the following best describes your child’s speech? Check all that apply.

- easy to understand
- difficult for others to understand
- difficult for parents to understand
- almost never understood by others
- different from other children of the same age

Which of the following best describes your child’s reaction to his/her speech?

- is easily frustrated when not understood
- does not seem aware of speech/communication problem
- has been teased about his/her speech
- tries to say sounds or words more clearly when asked
- Is successful in saying sounds or words more clearly when s/he tries

Does your child have difficulty producing certain sounds? _______ Yes _______ No
If “yes,” which ones?
______________________________________________________________________________

Does your child hesitate and/or repeat sounds or words? _______ Yes _______ No
Does your child “get stuck” when attempting to say a word? _______ Yes _______ No
Do you have concerns about your child’s voice? _______ Yes _______ No

Which of the following do you think your child understands? Please check all that apply.

- his/her own name
- names of body parts
- family names
- names of objects
- simple directions
- complex directions
- conversational speech

**Therapy History**

Has your child received speech/language therapy services? □ Yes □ No
Describe services provided ____________________________________________________________

Has your child received occupational therapy services? □ Yes □ No
Describe services provided ____________________________________________________________
Has the child received physical therapy services? □ Yes □ No
Describe services provided: ____________________________________________________________

Please complete all questions, even if your child is not currently in that therapy.

Is your child walking? □ Yes □ No
Is your child able to carry up to 5 lbs. of weight? □ Yes □ No
Does your child use any equipment or aides to walk? □ Yes □ No
Does your child have any gross motor concerns (i.e. rolling, crawling, sitting, walking with assistance, walking independently)? □ Yes □ No
If yes, please provide specific information:
___________________________________________________________________________________________

Is your child is physically limited? □ Yes □ No
If yes, please provide specific information on how he/she is frequently positioned (e.g. adapted seating, wheelchair, prone stander, on the floor, etc.):
___________________________________________________________________________________________

Does your child have any fine motor concerns (i.e. reaching, grasping, feeding, keyboard use/writing with pencil)? □ Yes □ No
If yes, please provide specific information: ____________________________________________________________

Which hand does your child prefer using? □ Right □ Left
Does your child have any limitations to the use of either hand? □ Yes □ No
If yes, please provide specific information: ____________________________________________________________

Is your child able to:
push a button on a computer keyboard? □ Yes □ No
use a computer mouse? □ Yes □ No
hold a fork or spoon without assistance? □ Yes □ No

Please list all previous speech/language therapy, occupational therapy, physical therapy, psychological or educational evaluations.

Communication Skills
How does your child currently communicate in the home? (e.g., requesting, commenting, asking/answering questions, clarifying and social exchange.) Please tell how the child uses the following ways to communicate and please list as many examples as possible.

Vocalizations/Verbalizations: (i.e. sounds; grunts; limited words/word approximations; phrases; complete sentences.) ____________________________________________________________

Reliable Yes/No Response: ____________________________________________________________
Gestures: ____________________________________________________________

Sign Language: ____________________________________________________________

**Pictures/Communication Boards/Overall Communication Strategies**

Has your child used the Picture Exchange Communication System (PECS)?  
☐ Yes  ☐ No

If yes, approximately how many symbols does the child independently use? (e.g. 10, 50, 100) __________

Where are the pictures kept? ________________________________________________________

How does the child access their pictures/communication board?  
☐ Independently  ☐ requires assistance

*Please bring any PECS symbols, books, communication boards or communicate devices your child currently uses or has previously used to their evaluation appointment.*

Has your child been using any communication systems?  
☐ Yes  ☐ No

What voice output systems has your child tried and with what success? (e.g. Communication Builder, Tech Talk; Go Talk, DynaVox Maestro, PRC /Vantage, iPad etc.): ______________________________

What activities or rewards (food or otherwise) can be used during this evaluation that will be motivating to your child? *Please bring a supply of “rewards” with you to the evaluation.* ______________________________

Describe your child’s preferred system(s) of communication/interaction: ______________________________

Percentage of time your child is understood: by familiar people: _______%  by unfamiliar people: _______%

How does your child interact with unfamiliar people? ______________________________

What does your child do when they are not understood? ______________________________

If your child is non-verbal, does he/she point or gesture to indicate wants and needs? ______________________________

Does your child attempt to imitate sounds or words?  ☐ Yes  ☐ No

Please check those areas in which the child finds a need to communicate:  
☐ home  ☐ community  ☐ telephone  ☐ medical/therapy appointments  
☐ school  ☐ telephone  ☐ medical/therapy appointments  
☐ work  ☐ medical/therapy appointments  
☐ other ______________________________________________________________________
Please check all that your child communicates with

☐ extended family  ☐ teachers  ☐ parents

☐ friends  ☐ classmates  ☐ children

☐ siblings  ☐ doctors  ☐ therapists

☐ other ____________________

Are there any hearing concerns for your child?  ☐ Yes  ☐ No

Does your child wear hearing aids or have a hearing loss?  ☐ Yes  ☐ No

What is the degree of hearing loss? ________________________________________________________________

Does your child have vision concerns (e.g., depth perception, visual attention to objects, following objects with their eyes, large print wording, etc.)? ________________________________________________________________

Does your child wear glasses/contacts?  ☐ Yes  ☐ No

Does your child have difficulty with any sensory input (touch, visual, noise)?  ☐ Yes  ☐ No

If yes, please provide specific information: __________________________________________________________

_____________________________________________________________________________________________

_____________________________________________________________________________________________

Name any medications that your child is currently taking: _______________________________________________

_____________________________________________________________________________________________

School History

Current Grade: ___________________________________________________________________

How often does your child attend classes? (Please choose one.)

☐ daily  ☐ 4 times per week  ☐ 2 times per week

Does your child’s developmental performance seem to interfere with his/her school performance?

Have teachers expressed any concerns about your child’s learning behavior?

If so please describe ________________________________________________________________

Is your child frequently absent from school? _____ If so, why? __________________________________________

How does your child feel about school and his or her teacher(s)? __________________________________________

Does your child receive special reading or language arts services? _____ If so, please explain:

_____________________________________________________________________________________________

_____________________________________________________________________________________________

Are there any special needs being addressed in the classroom?  ☐ Yes  ☐ No

If yes, please provide specific information: __________________________________________________________

_____________________________________________________________________________________________
Does your child currently have an:

Individual Family Service Plan (IFSP)?
- [ ] Yes
- [ ] No
- [ ] N/A

Individual Education Plan (IEP)?
- [ ] Yes
- [ ] No
- [ ] N/A

*If yes, please include report when submitting this questionnaire.*

Has a **Collaborative Problem Solving Assessment** been initiated or completed through the school?
- [ ] Yes
- [ ] No
- [ ] N/A

*If yes, please include report when submitting this questionnaire.*

Does your child receive the assistance of an aide in the classroom?
- [ ] Yes
- [ ] No
- [ ] N/A

If yes, please provide specific information:

__________________________________________________________________________________________

__________________________________________________________________________________________

How does your child communicate in the classroom setting?

__________________________________________________________________________________________

__________________________________________________________________________________________

If your child is in school, does he/she have difficulties learning to read or write?
- [ ] Yes
- [ ] No
- [ ] N/A

If yes, please provide specific information:

__________________________________________________________________________________________

__________________________________________________________________________________________

Please use the following space to provide any additional information you feel might be beneficial to the evaluation team.

__________________________________________________________________________________________

__________________________________________________________________________________________

__________________________________________________________________________________________

__________________________________________________________________________________________

__________________________________________________________________________________________

__________________________________________________________________________________________

Date questionnaire completed:

Name of individual completing questionnaire:

Relationship to child:

Please sign and date below and return pages 1 through 7. Please keep page 8 for your information/records.

*Signature of individual completing the questionnaire* 

*Date*