Speech and Language Abilities Associated with 16p11.2 Microdeletion Syndrome: Case Report

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Introduction

With advances in molecular genetic testing, newly identified genetic conditions may be implicated in developmental delay, including speech and language delay. The report describes the diagnostic characteristics of 16p11.2 microdeletion syndrome, and reviews the case of a boy (C.P.) with this diagnosis, with emphasis on his speech and language development. His medical history, behavior and speech and language abilities will be described and discussed.

Purpose

1) Describe clinical characteristics of 16p11.2 microdeletion syndrome.
2) Review the case of a child with 16p11.2 microdeletion syndrome and speech and language delay.
3) Understand the significance of speech and language delays associated with this genetic condition.

CHARACTERISTICS OF 16p11.2 MICRODELETION SYNDROME

Individuals with 16p11.2 microdeletion syndrome typically have developmental delay and intellectual disability. Additional characteristics include features of autism spectrum disorder (ASD) and delayed speech and language development, with greater delays in expressive skills than receptive language. IQ scores range from mild intellectual disability to normal (Hancock et al., 2010). Neuropsychological and/or developmental assessment and periodic reevaluation by a medical geneticist is recommended.

Congenital anomalies that have been found in individuals with 16p11.2 microdeletion syndrome include congenital diaphragmatic hernia, cleft palate, preaxial polydactyly, congenital heart disease, pyloric stenosis, cryptorchidism, clubfoot, and hypocalcemia. There is no particular pattern of physical anomalies that characterizes 16p11.2. Some individuals with the deletion have no identified physical, intellectual, or behavioral anomalies.

Clinical diagnosis of 16p11.2 microdeletion syndrome is based on clinical development, learning disabilities/intellectual disability, social impairments with or without a diagnosis of ASD and minor dysmorphic facial features without a consistent pattern.

Clinical and demographic findings in patients with 16p11.2 deletions

Table: Clinical and demographic findings in patients with 16p11.2 deletions

<table>
<thead>
<tr>
<th>Condition</th>
<th>Number of patients</th>
</tr>
</thead>
<tbody>
<tr>
<td>Male: Female</td>
<td>3:2</td>
</tr>
<tr>
<td>Mean age of diagnosis (years)</td>
<td>6 ± 1</td>
</tr>
<tr>
<td>Mean age of diagnosis (months)</td>
<td>54 ± 10</td>
</tr>
<tr>
<td>Delay in speech and language development</td>
<td>9 ± 2</td>
</tr>
<tr>
<td>Developmental delay</td>
<td>8 ± 2</td>
</tr>
<tr>
<td>Global developmental delay</td>
<td>7 ± 1</td>
</tr>
<tr>
<td>Eye accommodation problems</td>
<td>3/16</td>
</tr>
<tr>
<td>Congenital anomalies</td>
<td>5/16</td>
</tr>
<tr>
<td>Seizures</td>
<td>5/16</td>
</tr>
<tr>
<td>Birth defects</td>
<td>5/16</td>
</tr>
<tr>
<td>De novo/inherited</td>
<td>8/2</td>
</tr>
<tr>
<td>Male: female</td>
<td>3.2:1</td>
</tr>
</tbody>
</table>

Characteristics of 16p11.2 deletion

- Delayed gross motor development
- Delayed visual problem-solving skills
- Seizures
- Pyloric stenosis
- Congenital heart disease
- Pulmonary hypoplasia
- Persistent tachypnea
- Sleep apnea
- Congenital diaphragmatic hernia
- Cleft palate
- Clubfoot
- Cryptorchidism
- Preaxial polydactyly

C.P. is presently a seven-year-old male who has received comprehensive care in family medicine, medical genetics, and behavioral psychology. Verbal/language abilities were an area of weakness and fell in the lower average general intellectual functioning range with a full scale IQ = 87. Verbal/language abilities were low average general intellectual functioning with a full scale IQ = 87. Verbal/language abilities were an area of weakness and fell in the lower average general intellectual functioning range with a full scale IQ = 87. Verbal/language abilities were an area of weakness and fell in the lower average general intellectual functioning range with a full scale IQ = 87.

Speech-Language Re-Evaluation: Age 5 years

At age 5 years and 6 months, C.P. was re-evaluated for speech and language development. C.P. had been referred to speech therapy because of delays in speech and language development and later delays in speech, language, and cognitive development. Neuropsychological evaluation at age 6 years and 5 months revealed low average general intellectual functioning with a full scale IQ = 87. Verbal/language abilities were an area of weakness and fell in the lower average general intellectual functioning range with a full scale IQ = 87.

Language and Communication Assessment: Age 5 years

- C.P. demonstrated mild motor planning errors in connected speech. A phonetic inventory identified the following consonants: /t/, /b/, /m/.
- He was referred to audiology and speech-language pathology for further assessment.

Speech-Language Re-Evaluation: Age 5 years

At age 5 years and 6 months, C.P. was seen for speech therapy following his first author with therapy focused on remediation of sound errors and improved motor planning, particularly in connected speech. He was also referred to speech therapy in the school setting.

Behavioral Characteristics will be described.

In addition to growth delay and developmental delay, children with 16p11.2 deletion may also have behavioral and psychiatric problems. These include autism spectrum disorder and other neurodevelopmental conditions, such as attention-deficit hyperactivity disorder (ADHD), learning disabilities, and anxiety disorders.

References


This chapter describes the diagnostic characteristics of 16p11.2 deletion and speech and language development as a core feature and to consider referrals for developmental and medical evaluation in children who present with speech-language delays, including childhood apraxia of speech, and behavioral concerns.