This guidebook was developed by the Simons VIP Connect Study Team to help you learn important information about individuals with 16p11.2 duplication syndrome. Inside, you will find that we review everything from basic genetics and features of 16p11.2 duplication syndrome, to a description of clinical care and management considerations.

- Simons VIP Connect
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How Did We Collect All of this Information?

What is Simons VIP Connect?
Simons VIP Connect is a research project aimed at characterizing specific genetic diagnoses that have been associated with autism, such as differences in speech and language development, social interaction, and behavior.

Features of 16p11.2 Duplication

Thanks to the many 16p11.2 duplication families who have participated in the Simons Variation in Individuals Project (Simons VIP), we have learned a wealth of information about 16p11.2 duplication syndrome. This guidebook provides an in-depth summary of findings from the on-going Simons VIP study. We are excited to share this information with you and other newly-diagnosed families. Simons VIP research has helped us to better understand what to expect for children and young adults with 16p11.2 duplication syndrome. The success of this project can be attributed to the close partnership between families and researchers.
What is the Simons Variation in Individuals Project (Simons VIP)?

In 2010, the Simons Foundation began working to build a collaborative relationship between families and researchers to advance our understanding of 16p11.2 duplication syndrome. Families shared their medical histories and participated in different assessments, and researchers compiled this information and published what they’ve learned in medical journals. When families and researchers partner together like this, we are able to address questions quickly and give back to the 16p11.2 community. Families involved in Simons VIP Connect not only receive useful feedback by participating in research surveys, but they also are helping other families now and in the future.

Simons VIP Connect has been an important resource for connecting and supporting families who once felt alone after hearing about the diagnosis of 16p11.2. Now, the online community at Simons VIP Connect extends from our website to other social networks, like Facebook, where families from all over the globe support each other by sharing their stories and advice.

If you are not already a part of the online community, please visit our website at https://www.simonsvipconnect.org. There you can learn more about 16p11.2 duplication syndrome by participating in research, watching informational webinars, or asking an expert any questions that haven’t been answered in this guidebook.

Simons VIP Connect Research

In the Simons VIP study, families partner with researchers to share information and learn about the behavioral, psychological, physical, and medical features associated with different genetic changes. In particular, Simons VIP is interested in genetic changes known to cause autism and other neurodevelopmental disorders. Sharing information with Simons VIP Connect gives researchers access to an incredible repository of data, including your child’s de-identified medical information, which advances our knowledge about genetic causes of these disorders. Our model demonstrates the value of sharing information to create a resource that is accessible by researchers across multiple medical disciplines.
Definition of 16p11.2 Duplication

In every cell there is a copy of our DNA, which contains all of our genetic information. DNA is written in a code made up of four letters – A, C, T, G – the “spelling,” or order, of these letters makes up the instructions for everything our body does. Our DNA is very long so it needs to be coiled up into packages, or structures we call “chromosomes.”

![Diagram of DNA and chromosomes]

Humans have 46 chromosomes. We label the pairs of chromosomes 1 through 22, with the last pair being the sex chromosomes. These are made up of either two X chromosomes (female) or one X and one Y chromosome (male). Chromosomes are arranged into pairs because every person receives one half of the pair from their mother and the other half of the pair from their father.

DNA is broken up into individual, readable segments called “genes.” We believe that humans have about 20,000 genes, and each gene provides a set of instructions for the body to perform a specific job.

When a person has a 16p11.2 duplication, typically we find that a group of about 29 genes (~600,000 letters of DNA code) are repeated, or duplicated (some people may have more or less than this number). This means that one chromosome has the typical number of genes, while the other chromosome has extra information. Researchers now know that this specific section of chromosome 16 contains genes that play an important role in health, development, and brain function.
What is a Copy Number Variant (CNV)?
The 16p11.2 duplication is also often referred to as a Copy Number Variant, or CNV. A CNV is a difference in the number of copies of a particular section of genetic material. Since most people have two copies of their genetic material (as described above – one copy from mom and one copy from dad), a deletion (i.e., missing genetic information in a chromosome) or duplication (i.e., extra genetic information in a chromosome) is considered a CNV.
**Inheritance**

This duplication can happen one of two ways. In most families, the duplication is inherited; meaning that either mom or dad also has the 16p11.2 duplication and passed it on to their child. Simons VIP data shows that three-quarters (75%) are likely to be inherited. However, it can also occur de novo, meaning that the duplication is brand new in the family. Simons VIP data shows that one-quarter (25%) are thought to occur de novo. This means that 1 out of 4 children with a 16p11.2 duplication did not inherit it from mom or dad.

If a parent of a child with a 16p11.2 duplication is found to carry the duplication, then there is a 50% that any of his or her other children will may have the same duplication.

Since this duplicated genetic material can be passed down from a parent to their children, it is possible to test other family members to see if they have this same duplication. There are also tests that can be done before, during, or after pregnancy to find out if the next child will have the same 16p11.2 duplication a parent carries. Talk to your genetic counselor about these testing options.

When a child is found to have a 16p11.2 duplication, parents are often offered genetic testing.

If a parent is found to have a duplication, this means that any of his/her children had a 50% chance to have inherited it.

In this example, Dad has a 16p11.2 duplication (extra copy of 16p11.2 region shown in yellow). Of his four children, he passed the deletion to one son and one daughter, and his other son and daughter did not inherit the deletion.
How is a 16p11.2 Duplication Found?

Children diagnosed with 16p11.2 duplication syndrome have had genetic testing, as this is a genetic diagnosis. There are different types of genetic tests, and the ones we talk about here are able to detect a typical 16p11.2 duplication.

Genetic testing has improved over the past 10-15 years, and we are now able to identify many different types of genetic changes in people. We have genetic tests, including the microarray test, that look for many different genetic changes all at once. Microarray testing is often the first test ordered when a health care provider suspects that there may be a genetic reason for a child’s delays.

Another test, called Fluorescence In Situ Hybridization (FISH), may also be used to detect the 16p11.2 duplication. Since FISH testing is only targeted to one specific region/condition, some providers may order this testing to determine if other relatives also carry the specific genetic change that was identified in the family. However, the Simons VIP study team does not recommend that FISH testing be used to detect 16p11.2 duplications. This is because some labs have trouble clearly and reliably detecting the signal from extra genetic material, which may lead to an incorrect result. Instead, for families with 16p11.2 duplications, we would suggest that a “targeted microarray” be used to test family members.

*Slide 2: This project was made possible through the National Society of Genetic Counselors and a grant from the Audrey Heimler Special Project Award*
How Common are 16p11.2 CNVs?

A loss or gain of material (called a “copy number variant” or “CNV”) from 16p11.2 is increasingly recognized as one of the most common chromosome disorders. One out of a 100 children formally diagnosed with autism will have a 16p11.2 CNV (16p11.2 duplication or deletion), as demonstrated in the picture below. This makes the 16p11.2 CNV one of the most common genetic causes of autism. It is important to remember that not all children with 16p11.2 duplication will have autism.

Based on several research studies, we believe the prevalence of 16p11.2 CNVs are:

- 1/1900 people has a 16p11.2 duplication
- 1/2300 people has a 16p11.2 deletion

1 in 100 Individuals with a diagnosis of autism will have a 16p11.2 Duplication or Deletion
Most Common Features of 16p11.2 Duplication Syndrome

What Makes Something a “Syndrome?”

A syndrome is a recognizable pattern of features, signs and symptoms (such as medical, developmental, and behavioral concerns or characteristic physical findings) that occur together due to the same underlying cause. The characteristics of 16p11.2 duplication syndrome include a wide range of different developmental and behavioral concerns, as well as some specific medical conditions. Importantly, the syndrome can be quite variable from one person to the next, not everyone with the duplication will have the same abilities or challenges.

For example, some people with the 16p11.2 duplication had speech therapy in school, but never had any other medical problems. We would consider these people to be “mildly affected.” Other people with the duplication may have more significant learning and behavioral issues, or medical problems. The reason for such a great degree of variation among individuals who have the same (or a similar) duplication is still being studied. At this point, we do know that family background, environmental factors, and other genetic differences all contribute to how a person is affected by a 16p11.2 duplication.

While it is possible for someone with a 16p11.2 duplication to have no noticeable health or behavioral problems, this is not typical. The majority of people with 16p11.2 duplication syndrome have a combination of the features described in the sections that follow.
It can be helpful to know what range of medical, learning, and behavior issues can occur in people with 16p11.2 duplication syndrome, so that the correct supports are in place. The most common features fall into the categories of: developmental delay, behavior, growth patterns, and other medical issues.

**Differences in Development**

**Thinking and Learning Skills (Cognition)**

**Most children and adults with 16p11.2 duplication syndrome have trouble with learning and understanding.** Intelligence Quotient (IQ) is one measurement used to assess a person’s intelligence. Children with 16p11.2 duplication syndrome often have lower IQs than their siblings and parents who don’t have the duplication. Of course, there is still a wide range in IQ within the 16p11.2 duplication population. The average IQ in the general population is 100, with any number between 85 and 115 being considered average. Among people with the duplication, we believe the average IQ is about 26 points lower than their parents without the duplication.

We have worked with families whose children with 16p11.2 duplication syndrome have IQs above 100 and as low as 30. It is important to remember that an IQ score is just one measure of intelligence, and may not reflect a person’s social, adaptive, and other skills. We know that many people with the 16p11.2 duplication live and function independently, have meaningful relationships, and hold jobs.

A 2015 study of cognition in 270 people with the duplication shows that there is a large range of cognitive abilities in people who have the duplication. The variation observed in IQ score is not correlated with the presence of autism spectrum disorder or seizure status. This means that we do not think the presence of seizures, or a diagnosis of autism, will affect a person’s cognitive abilities.

**Speech and Language**

**Speech and language problems are the most common features seen in 16p11.2 duplication syndrome.** 80% of individuals with the syndrome have a diagnosed speech or language-related disorder. Language delay may be one of the first signs of 16p11.2 duplication that parents notice in their child. Families have told us that using sign language and communication devices can help with early communication skills.

**Motor Skills**

**Children with 16p11.2 duplication may have delays in achieving motor milestones.** 30% of individuals with the duplication are delayed at learning how to walk. The vast majority of children with a 16p11.2 duplication achieve major milestones such as sitting, crawling, and walking; however, these milestones are often achieved later
than expected. For example, a small number of children with a 16p11.2 duplication have been noted to not start walking until two years of age. The median age of onset for walking among children with the duplication is 18 months. Scientists believe that this is probably due to differences in the way their brains developed. **Low muscle tone** (hypotonia) is also frequently seen in infancy. Other signs of delayed motor skills include: clumsiness (dyspraxia), poor balance, poor posture, poor mobility, and difficulty grasping objects.

**Feeding Difficulties**

Trouble with feeding has been reported by some parents for their child’s first few weeks or months. While some children feed without any issues, others have trouble latching, sucking, and swallowing due to low muscle tone (hypotonia). These feeding difficulties can lead infants to have trouble gaining weight early in life. To help with this issue some babies have used a modified teat that is designed to help smaller babies get more milk. In more extreme cases, feeding tubes are used to make sure an infant is getting necessary nutrition.

**Behavior**

*Autism Spectrum Disorder (ASD)*

One in five individuals with the 16p11.2 duplication have a clinical diagnosis of Autism Spectrum Disorder (ASD); most people with 16p11.2 duplication syndrome do not have autism.

In large studies of people with autism, researchers have found that a small percentage of that population have a 16p11.2 duplication. This means, we know that 16p11.2 duplications can cause autism, but it is not the only known cause as there are many different genetic changes that have been identified in people with autism. In fact, even though autism is probably one of the most widely known diagnoses related to 16p11.2 duplication syndrome, the majority of people with a duplication are never diagnosed with autism. Research shows that about 20% of people with the duplication are given a diagnosis of autism spectrum disorder, whereas 80% of people with the duplication do not meet strict diagnostic criteria, but may have some autistic features.

It is much more common for a child with the duplication to exhibit some, but not all features of autism. One example of an “autistic-like” behavior is repetitive/restrictive behaviors (RRBs), meaning the child may like lining up items, or putting items in a specific order.

*Attention Deficit Hyperactivity Disorder (ADHD) and other behavioral diagnoses*

40.8% of individuals with the 16p11.2 duplication are found to have a behavior or psychological diagnosis that is **not** autism. ADHD is one of the most common behavior disorders diagnosed in
individuals with the 16p11.2 duplication. Some children with 16p11.2 duplication may use medications
to help increase attention span and reduce hyperactivity. Other diagnoses include anxiety, obsessive-
compulsive disorder (OCD), sleep disorders, and schizophrenia.

**Adult Onset Conditions**

Schizophrenia is a psychiatric condition that affects a person's ability to think clearly, manage emotions,
make decisions, and relate to others. Onset typically occurs in early adulthood. Schizophrenia has been
reported more frequently in individuals with 16p11.2 duplication syndrome than in the general
apopulation. In a 2015 research article, four individuals out of 270 people with a 16p11.2 duplication
were noted to have a diagnosis of schizophrenia; this is about 1.5% (the prevalence of schizophrenia in
the general population is around 1%). More research needs to be done on the risk of developing this
psychiatric illness, especially because many of the participants in Simons VIP Connect and other projects
are under age 18, which is prior to the typical age-of-onset for schizophrenia. Besides schizophrenia,
other psychiatric conditions, such as bipolar or depression, have been seen in individuals with the
16p11.2 duplication.

![Behavioral Diagnoses for Individuals with 16p11.2 Duplications](image)

*Other behavioral diagnoses
include, but are not limited to: schizophrenia, depression, bipolar disorder, or behavior
problems such as attention deficit hyperactivity disorder*
Growth Patterns

Decreased Head Size (microcephaly)

A smaller head size is common in people who have the 16p11.2 duplication. About 22% of individuals have microcephaly (a smaller-than-average head size), which typically develops over the first two years of life. In many cases, a smaller-than-average head size does not directly cause neurological problems, but has been associated with lower non-verbal IQ; this relationship is still being studied. Researchers have not found an association between smaller-than-average head size and seizures; this means the size of a person’s head does not influence the likelihood for seizures.

The 16p11.2 duplication affects brain size in a “dose-dependent” fashion, meaning that someone who has a 16p11.2 duplication may have a smaller head size than someone without the duplication. People with a 16p11.2 deletion (1 copy of this region) tend to have a larger-than-average head size.

Infant Head Size
BMI (Body Mass Index)

BMI is a measure of the weight for a person’s height. In people with the duplication, this measurement tends to be consistently below average throughout the lifespan. In individuals with the deletion, BMI tends to be higher than average.

Typical BMI development for boys and girls
Neurological Issues

Brain Structure and Function
As part of the initial Simons VIP in-person visits, individuals with 16p11.2 duplications had magnetic resonance imaging (MRI) to study their brains and compared them to their family members and to people who do not have the 16p11.2 duplication. This study demonstrated how individuals with a 16p11.2 duplication have an overall smaller brain size and shape. Particularly, this study showed how individuals with a 16p11.2 duplication have decreased intracranial volume, brain volume, gray matter, white matter, cerebellum, thalamus, hippocampus, amygdala, cortical thickness, and cortical surface area.

This study also demonstrated that a subset of the 86 16p11.2 duplication carriers had enlarged ventricles and cerebellar hypoplasia (13 [15.1%] and 10 [11.6%], respectively).

For more information on these areas of the brain, please visit this website: http://www.mayfieldclinic.com/PE-AnatBrain.htm

Seizures
Seizures are present in about 13.7% (or approximately 1 out of 7 individuals) with the 16p11.2 duplication. Focal epilepsy has been the most frequently observed epilepsy disorder in individuals with the duplication. All seizures are caused by abnormal electrical disturbances in the brain; focal or “partial” seizures occur when the electrical activity occurs in a limited area of the brain. A smaller percentage of individuals with 16p11.2 duplication have generalized seizures. This type of seizure affects both sides of the brain (cerebral hemispheres) and causes loss of consciousness. It is important to note that a “broad spectrum” of severity has been observed. Some seizures are severe and noticeable, while other seizures (such as absence seizures) may look like the child is “zoning-out” or day-dreaming. Because epilepsy is a relatively common neurological disorder observed in people with a 16p11.2 duplication, a referral should be made to a neurologist if any seizures are suspected. Some families find it useful to take pictures or videos of their child when he/she is having suspected seizure-like activity. Recordings of these episodes may help the neurologist to better understand and diagnose what your child is experiencing.
### Other Medical Problems

<table>
<thead>
<tr>
<th>OTHER MEDICAL PROBLEMS</th>
<th>HOW COMMON IS IT IN A PERSON WITH A 16P11.2 DUPLICATION?</th>
<th>DESCRIPTION OF HEALTH PROBLEM</th>
</tr>
</thead>
<tbody>
<tr>
<td>VISION PROBLEMS OR EYE ABNORMALITIES</td>
<td>15%</td>
<td>Near-sightedness, blurry vision, or the inability to focus both eyes on the same point</td>
</tr>
<tr>
<td>GASTROESOPHAGEAL REFLUX DISEASE (GERD)</td>
<td>10%</td>
<td>Damage to the lining of the esophagus due to stomach acid repeatedly being sent up into esophagus</td>
</tr>
<tr>
<td>SCOLIOSIS</td>
<td>8%</td>
<td>Abnormal curvature of the spine that develops over time</td>
</tr>
<tr>
<td>GENITAL MALFORMATIONS</td>
<td>6%</td>
<td>Differences in the growth / development of the male or female genitalia</td>
</tr>
<tr>
<td>CONGENITAL HEART DISEASE (atrial septal defects, ventricular septal defects, Shone’s complex, tetralogy of Fallot)</td>
<td>4%</td>
<td>Heart problems present at birth are present in 1% of all babies, so it is not surprising that heart problems have been noticed in babies who also have 16p11.2 duplication. However, the chance is slightly higher than in the general population</td>
</tr>
<tr>
<td>DECREASED HEARING</td>
<td>4%</td>
<td>Partial hearing loss</td>
</tr>
<tr>
<td>ASTHMA</td>
<td>4%</td>
<td>Trouble breathing, usually initiated by physical activity</td>
</tr>
<tr>
<td>OROFACIAL</td>
<td>2%</td>
<td>A problem with the mouth or face, such as a hole in the roof of the mouth (cleft palate) or a blockage in the nasal passage</td>
</tr>
</tbody>
</table>
Evaluations Following Initial Diagnosis

At this point in time, there are not published guidelines for evaluation immediately following diagnosis. Management is largely symptom-based. These are only suggestions based on symptoms of individuals with 16p11.2 duplication. Parents of individuals with 16p11.2 duplication should consider:

1. Regular Check-Ins with Pediatrician
   
   a. Measure height and weight at every visit with the pediatrician to monitor growth and BMI
      
      i. Screening for malnutrition in who are underweight

2. Consider consultation with a neurologist and EEG testing if family or physician suspects seizures

3. In patients with spinal curvature, x-ray of the spine to evaluate for vertebral anomalies

4. Consider evaluation and echocardiogram by a cardiologist if a heart murmur is identified
Ongoing Follow Up and Management

*There are no guidelines for management of individuals with 16p11.2 duplication. These are only suggestions based on symptoms of individuals with 16p11.2 duplication. Parents of individuals with 16p11.2 duplication should consider:

1. Early diagnosis and provision of therapies facilitates the best outcome. Referral to other appropriate medical specialists is recommended based on specific symptoms or signs. Specialists may include a developmental/behavioral pediatrician, pediatric neurologist, and/or medical geneticist.

2. Due to the high incidence of neurodevelopmental disability, referral to a clinical psychologist for full neuropsychological and/or developmental assessment for diagnostic differential and treatment recommendations is strongly suggested. Interventions may include speech and language therapy, occupational therapy, and physical therapy. Because of the high incidence of expressive language delays, speech therapy and augmentative and assistive means of communication should be considered early. For families in the UK, these assessments would usually be carried out by a child’s neurodisability team.

3. Behavioral, social, and educational interventions for individuals with neurodevelopmental disabilities, including autism, are also appropriate. Guidelines for management of individuals with autism are available from the American Academy of Pediatrics.

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Different Duplications, Different Groups

Not every 16p11.2 duplication is the same, therefore it is important to understand the different duplications that can occur. There are different groups used to describe 16p11.2 duplications, based on the location, amount of duplicated genetic material, and number of genes.

**Group 1** has the “typical,” or most common, 16p11.2 duplication. This guidebook is tailored to individuals with a group 1 duplication.

**Groups 2** have duplications that are in a slightly different location than in group 1. These groups do not overlap. Group 2 duplications are found closer to the end of Chromosome 16; these are called “distal” 16p11.2 duplications.

**Group 3** is indicated in green. This duplication comprises genetic material from both groups 1 and 2.
Living with 16p11.2 Duplication Syndrome

Growing up
As children grow up, it may feel like the gap between your child with the 16p11.2 duplication and your other children is growing or that your child isn’t able to keep up with peers. The gap in academic skills between a child and his/her typical peers tends to widen over time. Children with the duplication are often still learning and achieving goals at school, but it may be at their own pace. It’s important to talk with your child’s teacher about developing an Individualized Education Program (IEP) that meets your child’s learning style and needs, but also challenges him or her.

Services
Typically, there are several different kinds of professionals and services involved with the care of a child with 16p11.2 duplication syndrome. The services needed depend on the features that your child is showing. You are going to play an important role in directing, organizing, and advocating for your child’s care. One or more of the following professionals may be involved in taking care of your child:

Genetic Counselors and Geneticists work with families with many different types of genetic diagnoses. A geneticist may help with the initial diagnosis, medical assessment, and coordination of appropriate referrals based on what we know about a child’s medical concerns related to the diagnosis.

Genetic counselors can educate families about the 16p11.2 duplication and discuss how the diagnosis affects their family. They can also coordinate genetic testing for family members and identify appropriate support resources. To locate a genetic counselor near you, visit www.nsgc.org and click “Find a Genetic Counselor.”

Pediatric Neurologists will be involved if there are concerns that seizures are occurring or if other neurological issues are present. If your child is having seizures, a neurologist can monitor and manage them.

Occupational Therapists work with people to help improve their ability to perform everyday tasks. They can evaluate the impact of the 16p11.2 duplication on daily activities at home, school, or work. For example, many children have trouble with their fine motor skills. Children with 16p11.2 duplication may struggle with handwriting, using scissors, turning pages, or using a computer. An occupational therapist can work with your child to come up with a personalized plan to improve their skills in these areas.

Physical Therapists can evaluate and provide therapy to improve motor skills and coordination. Therapy can include stretches, exercises, and body movements that increase a person’s motor abilities.

Nutritionists can help children manage their weight. Having 16p11.2 duplication syndrome does not mean someone will be underweight, but it may take more work for them to control their weight.
nutritionist is a helpful professional to meet a family’s needs for developing the right eating and weight management plan for children with 16p11.2 duplications.

**Developmental Pediatricians** are physicians who specialize in developmental and behavioral disorders in children, including autism, developmental delay, and intellectual disability. They can recommend appropriate medical treatments and behavioral interventions, and they can provide information about school and intervention services in your local area.

**Psychiatrists** are experts in the diagnosis and treatment of ADHD, obsessive compulsive disorder, schizophrenia and other mental health conditions. While there is no cure for these disorders, psychiatrists can prescribe and monitor medications to treat psychiatric symptoms.

**Psychologists and Social Workers** help children and adults emotionally process feelings and challenges they face in everyday life. Psychologists can diagnose and treat a wide range of emotional and behavioral problems.

**Primary Care Physicians** (PCP) serve as the “headquarters” for a child’s overall medical care. A child with a 16p11.2 duplication will still go for wellness check-ups every year, just like with any other child. In addition, a PCP can coordinate specialty referrals and make sure that any necessary lab tests are completed.

**Applied Behavior Analysis (ABA)** is a field of psychology that focuses on understanding behavior and how it is affected by a child’s environment, including interactions with other people. ABA-based techniques are used to teach a range of academic, social, communicative, motor, and adaptive skills. A few studies have shown ABA to be a helpful intervention for some children with ASD.

**Speech and Language Pathologists (SLPs)** Many individuals with 16p11.2 duplication syndrome work with a SLP. SLPs help children and adults with a variety of communication, reading, or swallowing problems.

**Speech Disorders**

- Articulation - the way we say our speech sounds
- Phonology - the speech patterns we use
- Apraxia - difficulty planning and coordinating the movements needed to make speech sounds
- Fluency - stuttering
- Voice - problems with the way the voice sounds, such as hoarseness

**Language Disorders**

- Receptive Language - difficulty understanding language
- Expressive Language - difficulty using language
- Pragmatic Language - social communication; the way we speak to each other
Other Disorders

- Deafness/Hearing Loss - loss of hearing; therapy includes developing lip-reading, speech, and/or alternative communication systems
- Oral-Motor Disorders - weak tongue and/or lip muscles
- Swallowing/Feeding Disorders - difficulty chewing and/or swallowing

There are several ways to find an SLP if you are concerned about your child's communication skills. Your school/school district should have a certified SLP that can observe and/or test your child for speech and language difficulties. Your local children's hospital will also have certified SLPs on staff. You will find them in departments such as: Department of Hearing and Speech, Clinic for Communication Disorders, or Developmental Clinic. Your family practitioner can also recommend an SLP. The American Speech-Language-Hearing Association (ASHA) provides a search engine to help you find a local, certified SLP in your area: [http://www.asha.org/findpro/](http://www.asha.org/findpro/)

(Information from [www.superduperinc.com](http://www.superduperinc.com))

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*If a child has been diagnosed with a speech or motor delay, he or she may be eligible to receive early intervention services. The American Academy of Pediatrics says that “early intensive behavioral and educational intervention can make a significant positive impact on long-term outcomes.” Early intervention can include many different types of therapies.*
What to Tell Teachers

Children with special learning and behavioral needs often receive an Individualized Education Program (IEP) through their school to help them reach their full learning potential. As mentioned, most children with a 16p11.2 duplication do not have a diagnosis of autism. We do know, however, that children with a 16p11.2 duplication often have some degree of intellectual disability or delay in development. It’s important to inform your child’s teachers about your child’s needs. It may be helpful to give them a copy of this guidebook so they can better understand 16p11.2 duplication syndrome.

A request for the school district to conduct a comprehensive intellectual and academic assessment and to perform testing to identify strengths/weaknesses is a good starting point. Your child’s progress should be monitored and he/she should receive an IEP and/or curricular modifications, if available. Having a tutor or aide to encourage a child and reinforce skills can be helpful for students who are feeling overwhelmed.

Some children participate in a typical classroom while others may require “pull-out” services. Other children may benefit most from participation in a special education classroom. Picking the right school setting for your child is an important decision that should involve you, your child’s teachers, administrators, school psychologist, and therapists.

Adaptive Technologies

There are numerous technologies that can aide a child with 16p11.2 duplication syndrome with learning and language skills. The best approach to finding out what kind of adaptive technologies can best help your child is to have an Assistive Technology (AT) or Augmentative /Alternative Communication (AAC) evaluation from a skilled clinician.

When selecting a speech device, it’s important to take the following points into account:

- the student’s interest in and comfort level with the technology
- the student’s ease in learning about and using the technology
- the degree to which the technology “taps” into the student’s strengths
- the extent to which the student is able to use the technology independently and “troubleshoot” as necessary
- the effectiveness of the technology in compensating for specific difficulties as compared to alternative strategies

(Information retrieved from http://www.greatschools.org/pdfs/e_guide_at.pdf)
Tablets/iPads (mobile computer) can now be used for adaptive technology. While the Simons VIP Connect researchers and clinicians do not endorse any specific assistive devices, it may be helpful to watch one of our webinars on selecting an iPad at https://simonsvipconnect.org/en/about-cnvs/our-webinars/500.

In this webinar, Kelly A. Johnson, PhD of the UW Autism Center describes important qualities to consider when selecting an iPad and applications (apps) to use as a communication device for your child. Before buying an iPad it’s important to consider the iPad size that will best fit your child’s need. For example, if he/she has poor hearing, a larger device will have larger speakers. The amount of storage needed, types of accessories, such as a stylus, insurance, a protective case, built in features, and of course cost are all things to take into consideration when picking out an iPad. Once you have an iPad for your child, in order for him/her to get the full benefit you will both need expert guidance on which apps to use to attain goals.

Apps can be used for a variety of functions such as communication, social interaction, education, and even distraction during medical procedures. When choosing apps to use with your child, it’s important to remember that the ones that cost money are not necessarily the most useful. While there are many apps out there catering to children with special needs, mainstream apps can be helpful as well. The iPad itself has a lot of useful tools built in such as video chat and the internet. If a child is using an iPad for communication, it’s important to know that it should only be used by him or her. The iPad is working as your child’s voice, so nobody else in the family should be using it. Lastly, while an iPad can be an excellent adaptive technology, it is not the only useful technology out there.

Learn more about the latest and most liked apps at:

Acknowledgements

There are many people to thank in a project as large as this one. First and foremost, we want to acknowledge the sponsor of the Simons Variation in Individuals Project and Simons VIP Connect, the Simons Foundation Autism Research Initiative. Their commitment to improving the understanding, diagnosis and treatment of autism spectrum disorders has brought together a team of experts to lead the way in understanding 16p11.2 duplications and other genetic changes.

We must also thank the entire Simons VIP study team – including all of the investigators, study coordinators, project managers, and administrators, as well as our website developer, Patient Crossroads, and our data coordinating center, Prometheus Research – who have dedicated many hours to meeting with families and making sure the project runs smoothly. Their work, along with other collaborators, has led to numerous publications about 16p11.2 (listed below).

Finally, we owe a huge debt of gratitude to the many families who have participated in the Simons VIP study. Since 2010, over 160 individuals with the 16p11.2 duplication have participated in this research project, with many families still contributing updates to this day! This guidebook has been made possible thanks to the time, energy and information they have committed to sharing with us.

A special thank you is extended to our 2015 summer intern, Elly Brokamp, for developing the initial draft of this comprehensive guide, as well as to our 2016 summer intern, Nate Hassel, for making updates specific for 16p11.2 duplications. Thank you for your contribution; your work has had a tremendous impact.
## Resources and References

### Resources

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<tr>
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<th>VIP APPROVED WEB RESOURCES</th>
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<td>1</td>
<td>The Simons VIP Connect website <a href="https://simonsvipconnect.org/">https://simonsvipconnect.org/</a></td>
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<td>5</td>
<td>Genetic and Rare Diseases Information Center – 16p11.2 Duplication <a href="https://rarediseases.info.nih.gov/gard/12388/16p112-duplication/resources/1">https://rarediseases.info.nih.gov/gard/12388/16p112-duplication/resources/1</a></td>
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## Resources and References

### Articles Using Simons VIP Data

<table>
<thead>
<tr>
<th>YEAR PUBLISHED</th>
<th>TITLE</th>
<th>AUTHORS</th>
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<tbody>
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<td>2012</td>
<td>Simons Variation in Individuals Project (Simons VIP): A genetics-first approach to studying autism spectrum and related neurodevelopmental disorders</td>
<td>The Simons VIP Consortium</td>
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<td>Qureshi, et al.</td>
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<td><a href="http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4138332/">http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4138332/</a></td>
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<td>2015</td>
<td>Auditory Evoked M100 Response Latency Is Delayed in Children with 16p11.2 Deletion but not 16p11.2 Duplication</td>
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<td>Reciprocal white matter alterations due to 16p11.2 chromosomal deletions versus duplications</td>
<td>Chang, et al.</td>
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<td>2016</td>
<td>Deletion and duplication of 16p11.2 are associated with opposing effects on visual evoked potential amplitude</td>
<td>Leblanc, et al.</td>
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### Other Articles about 16p11.2 Duplication Syndrome

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<tr>
<th>YEAR PUBLISHED</th>
<th>TITLE</th>
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<tr>
<td>2009</td>
<td>Microduplications of 16p11.2 are associated with schizophrenia</td>
<td>McCarthy, et al.</td>
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<td>2010</td>
<td>Expanding the clinical spectrum of the 16p11.2 chromosomal rearrangements: three patients with syringomyelia</td>
<td>Schaaf, et al.</td>
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<td>2010</td>
<td>Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioral problems, dysmorphism, epilepsy, and abnormal head size</td>
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<td>2010</td>
<td>Speech delays and behavioral problems are the predominant features in individuals with developmental delays and 16p11.2 microdeletions and microduplications</td>
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<td>2015</td>
<td>Modulation of mu attenuation to social stimuli in children and adults with 16p11.2 deletions and duplications</td>
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