16p11.2 Duplication Syndrome Guidebook
This guidebook was developed by the Simons Searchlight study team to help you learn important information about people with a 16p11.2 duplication syndrome.

Inside, you will find a review of everything from basic genetics and features of 16p11.2 duplication syndrome to clinical care and management considerations.

– Simons Searchlight
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How Did We Collect All of This Information?

Simons Searchlight is a research project aimed at characterizing genetic changes that have been associated with autism and other neurodevelopmental conditions, such as differences in social interaction, behavior and cognitive skills.

People with a 16p11.2 duplication will have differences in:

Social Interactions

Cognitive Skills

Behaviors

Thanks to the many 16p11.2 duplication families who have participated in Simons Searchlight (formerly Simons Variation in Individuals Project), we have learned a wealth of information about 16p11.2 duplication syndrome. This guidebook provides an in-depth summary of findings from the ongoing Simons Searchlight study. The data you see here is from you. We are glad we can share this information with you and other newly diagnosed families. Simons Searchlight research has helped us 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 Simons Searchlight?

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 tests, and researchers compiled this information and published what they had learned in medical journals. When families and researchers partner like this, we are able to answer questions quickly and give back to the 16p11.2 community. Families involved in Simons Searchlight not only receive useful feedback by participating in research, but they also are helping other families now and in the future.

Simons Searchlight has been an important resource for connecting and supporting families who once felt alone after receiving a genetic diagnosis such as 16p11.2 duplication. Now the online community at Simons Searchlight 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 Searchlight online community, please visit our website at www.simonssearchlight.org/. There you can learn more about 16p11.2 duplication syndrome by watching informational webinars, participating in research, or asking an expert any questions that this guidebook hasn’t answered.

Simons Searchlight Research

In the Simons Searchlight study, families partner with researchers to share information and learn about the behavioral, psychological, physical, and medical features associated with certain genetic changes. In particular, Simons Searchlight is interested in genetic changes known to cause autism and other neurodevelopmental conditions. Sharing information with Simons Searchlight helps researchers build a vast repository of data, including de-identified medical information from you or your child. This advances our knowledge about genetic causes of these conditions. Our resource is used by researchers across multiple medical disciplines.
**Definition of a 16p11.2 Duplication**

The human body is made up of millions of cells. In every cell there is a copy of our DNA, which contains all of our genetic information. DNA is written in a code consisting of four base “letters” — A, C, T, G — that the body reads like sentences. The “spelling,” or order, of these letters is important to give the instructions for everything our body needs to function. Our DNA sentences are very long, so the DNA needs to be coiled up into packages, or structures we call chromosomes.

Humans have 46 chromosomes: 23 chromosomes from mom and 23 chromosomes from dad. This is why most of us look like both our parents. 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).

Humans have more than 20,000 genes. Each chromosome has a few hundred to thousands of genes. Each gene provides a set of instructions to make the building blocks of the human body.

When a person has a 16p11.2 duplication, typically we find that a group of about 29 genes (~600,000 letters of DNA code) is duplicated. This means that one chromosome has the typical number of genes, while the other chromosome has two copies of the same information for a total of three copies of this set of genes on the two chromosome 16s. Researchers now know that this
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 referred to as a copy number variant, or CNV. A CNV is a difference in the amount of genetic material. Since most people have two copies of their genetic material (one from mom and one from dad), a duplication (extra genetic information in a chromosome) is considered a CNV.

The picture above shows different genes in blocks of colors along one chromosome. A duplication is an extra set of these blocks. Someone with a 16p11.2 duplication will have one chromosome with two sets of genes in the duplicated section and one chromosome with one set.
Inheritance

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

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

If a parent is found to have a duplication, it means that any of their children have a 50% chance of inheriting it.

In the example at left, dad has a 16p11.2 duplication (shown in yellow). Of his four children, one son and one daughter inherited the duplication, and his other son and daughter did not inherit it.

Since this duplicated genetic material can be passed down from a parent to their children, if one of the parents has the duplication, it is possible that other family members, including aunts, uncles, and grandparents, have the duplication as well. There are tests that can be done before, during, or after pregnancy with the next child to find out if this child will have the 16p11.2 duplication that the parent carries. Talk to your genetic counselor about these testing options.
How Is a 16p11.2 Duplication Found?

Children diagnosed with 16p11.2 duplication syndrome have had genetic testing. 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 20 years, and we are now able to identify many types of genetic changes in people. Genetic tests now include a chromosome microarray test, which looks for copy number variants across a person’s chromosomes and can look for many different copy number variants at the same time. A chromosome microarray test is often the first test ordered when a healthcare provider suspects that there may be a genetic reason for a child’s issues.

Another test called fluorescence in situ hybridization (FISH) can also detect a 16p11.2 duplication. FISH testing is only targeted to one specific region/condition, so some providers may order this testing to determine if other relatives too carry the specific genetic change that was identified in the family. However, the Simons Searchlight study team does not recommend that FISH testing be used to detect 16p11.2 duplications. 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 chromosome microarray” be used to test family members.

How Common Are 16p11.2 CNVs?

A gain of material in 16p11.2 is one of the most common chromosome conditions. Several research studies indicate, that about one in 1,900 people in the general population have a 16p11.2 duplication.

About 1 in 200 people with autism has a 16p11.2 duplication. However, it is important to remember that not all children with a 16p11.2 duplication will have autism. Many people with the duplication are neurotypical.
Different Duplications, Different Groups

Not everyone with a 16p11.2 duplication is the same. Different 16p11.2 duplications have different groups, according to the location and the amount of genetic material duplicated.

**Group 1** has the “typical,” or most common, 16p11.2 duplication (shown in dark red below). *This guidebook is tailored to people with a group 1 duplication.*

**Group 2** has duplications that do not overlap with group 1 and are found closer to the end of chromosome 16. These are called distal 16p11.2 duplications. Group 2 distal 16p11.2 duplications are further grouped by size and location. Group 2a is a larger duplication (shown in blue), and group 2b is a smaller duplication on the distal edge of the group 2 region (shown in bright red).

**Group 3** is shown in green. This duplication is larger and includes genetic material from both groups 1 and 2.

*Image courtesy of Unique. More information on duplication groups 2 and 3 may be found in Unique’s microduplication guidebook.*
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 developmental and behavioral concerns, as well as some specific medical conditions. Importantly, the syndrome can be quite variable from one person to the next, and not everyone with the duplication will have the same abilities or challenges.

For example, some adults with the 16p11.2 duplication had speech therapy in school but have never had any other medical problems. Other people with the duplication may have more significant learning and behavioral issues or medical problems. The reasons for the variation among people who have the same (or a similar) duplication are still being studied. At this point, we know that several factors contribute to how a person is affected by a 16p11.2 duplication, such as other genes in the body and environmental factors.

It can be helpful to know what range of medical, learning, and behavioral issues can occur in children with 16p11.2 duplication syndrome, so that the correct supports are in place. The most common features fall into the categories of development, growth patterns, neurological issues, and other medical issues, which we discuss below.
Differences in Development

Thinking and Learning Skills (Cognition)
Many children with the 16p11.2 duplication syndrome have difficulties with learning.

Measures of cognitive ability are often lower in children with the duplication than in their family members without the duplication. Nearly half do not outgrow their learning delays. If learning delays are present across several areas of functioning and the child does not catch up at elementary school age, they may receive a diagnosis of intellectual disability and require special education classes and supports for daily living.

Adults with a 16p11.2 duplication who were discovered through family testing provide us with a different picture. They show us that the effect of the duplication varies widely and that above average cognitive ability is possible. This indicates that many children with the duplication will be able to live and function independently as adults. Careful reassessment is recommended for every child at major transition points during the school years and before graduation, to understand their need for supports as they get older.

Speech and Language
Speech delays are common in 16p11.2 duplication syndrome.

A majority (88%) of children with the duplication were able to speak by age 2½ according to research in Simons Searchlight, but one in three has difficulties speaking or understanding others when the language becomes more complex. When a child has difficulties with language but not with other kinds of learning or problem-solving, they may be diagnosed with language disorder, sometimes called specific language impairment. Families have told us that using sign language and communication devices can help children with language impairment improve their early communication skills. As the child grows, it is important for school providers to monitor for learning disabilities in reading and writing. These commonly follow language impairment and require special education services and targeted approaches to teaching reading.
**Motor Skills**

Children with a 16p11.2 duplication often have delays in motor coordination.

Children with the duplication may reach their “firsts,” such as first steps, a little later than expected. And motor development may continue to be delayed as the child grows. For example, children with a 16p11.2 duplication in Simons Searchlight tend to walk independently around 18 months, and nearly half are diagnosed with developmental coordination disorder. Researchers believe that this is due to differences in the way the brain develops. Low muscle tone, or hypotonia, is also common, even in infancy. Other problems with motor skills include difficulties in balance, posture, and grasping objects, and also clumsiness and motor planning problems, or dyspraxia. Physical therapy may be recommended.

**Feeding Difficulties**

Some parents have reported trouble with feeding during 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 problems can make it difficult for infants to gain weight early in life. To help with this issue, some babies have used a modified nipple on the bottle that is designed to help smaller babies get more milk. In extreme cases, feeding tubes are used to make sure an infant gets the necessary nutrition.
Behavior

Autism Spectrum Disorder

One in five people with the 16p11.2 duplication has a clinical diagnosis of autism.

In large studies of people with autism, researchers have found that a small percentage have a 16p11.2 duplication. This means that we know 16p11.2 duplications can cause autism — but it is not the only known genetic cause. Many other genetic changes have been identified in people with autism. Even though autism is 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.

More commonly, a child with the duplication will have some behaviors that are similar to those seen in autism, such as restricted and repetitive behaviors (RRBs). For example, the child may feel most comfortable doing only certain activities over and over, or may be intensely interested in one television show or activity. If a parent wonders if their child has autism, an evaluation is recommended as early as possible for a careful differential diagnosis, to make sure that therapies and school services will be a good fit for them.

Attention Deficit Hyperactivity Disorder (ADHD) and Other Behavioral Diagnoses

Many children with a 16p11.2 duplication have other behavioral and emotional issues, such as anxiety and difficulties with attention.

ADHD is one of the most common behavior disorders reported in children with a 16p11.2 duplication. About one in three people with a 16p11.2 duplication in Simons Searchlight reports that they use medications for behavior or mood, for example, to help increase attention span and reduce hyperactivity.
The graph below is a summary of clinical diagnoses in 62 children with a 16p11.2 duplication in the Simons Searchlight study.

**Developmental and behavioral diagnoses in children with a 16p11.2 duplication**

- Autism
- Language
- Speech Sound
- Learning Disorder
- Intellectual Disability
- Developmental Coordination Disorder
- ADHD
- Anxiety

**Adult-Onset Conditions**

The most common problems experienced by adults with a 16p11.2 duplication are anxiety and depression.

Besides anxiety and depression, other conditions seen in adults include obsessive-compulsive disorder and bipolar disorder. Earlier studies of 16p11.2 copy number variations suggested an increased risk of schizophrenia in adults (1.5% of adults with a variation vs. 1% in the general population). Schizophrenia affects a person’s ability to think and perceive their environment clearly. More research needs to be done on the risk of developing this condition, especially because many participants in Simons Searchlight and other studies are under age 18, which is younger than the typical age of onset for schizophrenia.
The graph below is a summary of clinical diagnoses in 41 adults with a 16p11.2 duplication in the Simons Searchlight study.

### Developmental and mental health diagnoses in adults with a 16p11.2 duplication

- Autism
- Intellectual Disability
- ADHD
- Anxiety
- Mood Disorder

### Growth Patterns

**Decreased Head Size (microcephaly)**

About one in five children with a 16p11.2 duplication has a smaller-than-average head, which typically is seen in the first two years of life. There is a possibility of seizures and lower cognitive ability in children who have microcephaly.

**BMI (Body Mass Index)**

**BMI tends to be below average for people with a 16p11.2 duplication.**

BMI is a measure of a person’s weight (kilograms) relative to their height (meters). You may find it helpful to talk with a dietitian about how to support your child in maintaining a healthy weight.

\[
BMI = \frac{\text{Your Weight}}{(\text{Your Height})^2}
\]
Neurological Issues

Neurological Exam Findings
In neurological exams performed in clinic in Simons Searchlight, the most common findings in people with a 16p11.2 duplication included low muscle tone and weakness, underactive or overactive reflexes (a test of nerve impulses), and motor tremor (slight shaking or trembling of the hands). These problems mean that it’s important for a child to have occupational and physical therapy evaluations.

Brain Structure and Function
As part of the initial Simons Searchlight study, people with a 16p11.2 duplication had magnetic resonance imaging (MRI) scans to study their brain structure. Their scans were compared with scans from their family members and people who do not have a 16p11.2 duplication. This study demonstrated that people with a 16p11.2 duplication have an overall smaller-than-average brain size, and decreased brain volume in all areas of the brain.

Seizures
About one in five people with a 16p11.2 duplication has seizures.

The type of epilepsy most commonly seen in people with a duplication is called focal epilepsy. All seizures are caused by abnormal electrical activity in the brain. Focal, or partial, seizures occur when the abnormal activity occurs in a limited area of the brain. Some people with a 16p11.2 duplication also have generalized seizures, which affect both sides of the brain. Different seizure types may occur in the same person. Some seizures are severe and noticeable, while other seizures may look like the child is “zoning out” or daydreaming. If you suspect your child is having seizures, ask your healthcare provider whether they should be evaluated by a neurologist or tested with an electroencephalogram (EEG). Some families find it useful to take pictures or videos of their child during seizure-like activity. These recordings may help the neurologist understand and diagnose what your child is experiencing.
**Other Medical Problems**

Other medical issues that are found are shown in the table and graph below.

This information was gathered from 141 people with a 16p11.2 duplication in Simons Searchlight. The most frequent problems reported were gastrointestinal problems (such as heartburn), and bone abnormalities such as scoliosis, which a healthcare provider should monitor as the child grows. Children with a duplication may also be prone to ear infections and respiratory infections. Other less common medical issues include vision problems such as strabismus (when the eyes do not align properly), and deafness.

<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 issue</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gastrointestinal (GI) problems</td>
<td>44%</td>
<td>Gastroesophageal reflux (heartburn), constipation, diarrhea</td>
</tr>
<tr>
<td>Bone abnormalities</td>
<td>26%</td>
<td>Scoliosis, the most common abnormality, is curvature of the spine that develops over time, often in adolescence. Other abnormalities may be seen in fingers, feet, or the chest.</td>
</tr>
<tr>
<td>Urogenital malformations</td>
<td>24%</td>
<td>Differences in the growth/development of the male or female genitalia, kidneys, or urinary system.</td>
</tr>
<tr>
<td>Heart conditions</td>
<td>25%</td>
<td>Including atrial septal defects, ventricular septal defects, Shone’s complex, and tetralogy of Fallot. Heart conditions are seen in 1% of all babies at birth, but they are more common in babies who have a 16p11.2 duplication.</td>
</tr>
</tbody>
</table>
Other medical problems in individuals with a 16p11.2 duplication

- GI Issues: 50%
- Bone Abnormalities: 25%
- Urogenital Malformations: 20%
- Heart Conditions: 15%
Evaluations After Initial Diagnosis

There are not now any published guidelines for what evaluations should be done immediately after diagnosis. What tests should be ordered mainly depends on the child’s symptoms. The recommendations below are based on symptoms common in people with a 16p11.2 duplication.

1. Regular check-ins with pediatrician
   a. Measure height and weight at every visit with the pediatrician to monitor growth and BMI

2. If you or a healthcare provider suspects seizures, consider consultation with a neurologist and EEG testing.

3. If a physical exam suggests the child may have scoliosis or other bone abnormalities, consider having X-rays done.

4. If a heart murmur is found, consider evaluation and an echocardiogram by a cardiologist.
Ongoing Follow-Up and Management\textsuperscript{1}

The following advice is based on symptoms of people with a 16p11.2 duplication.

1. Early intervention can lead to improved outcomes. Depending on symptoms, the child should be referred to medical specialists such as a developmental/behavioral pediatrician, pediatric neurologist, and/or medical geneticist.

2. Many children with a 16p11.2 duplication have neurodevelopmental disabilities. So referral to a clinical psychologist or developmental pediatrician is suggested. Children under age 6 should have psychological and neuropsychological assessments every year. Children aged 6 and older should have a psychological evaluation every three years and at major transition points. They may benefit from speech and language therapy, occupational therapy, and physical therapy. Because expressive language delays are common, speech therapy and other help with communication should be considered early. For families in the UK, these assessments would usually be carried out by a child’s neurodisability team.

3. Children with a 16p11.2 duplication, including those with autism, may also benefit from behavioral, social, and educational interventions. Guidelines for ongoing care for children with autism are available from the American Academy of Pediatrics.

Living with 16p11.2 Duplication Syndrome

Growing Up

Over time, you may feel as if there is a gap between your child with the 16p11.2 duplication and your other children, or that your child isn’t able to keep up with peers. The gap in academic skills between a child and their typical peers tends to widen as they grow up. Children with the duplication often still learn and achieve 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 them.

Services

Most children with 16p11.2 duplication syndrome receive services from several kinds of professionals. The services needed depend on your child’s symptoms. 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 help you care for your child:

**Genetic counselors and geneticists** work with families with many different genetic diagnoses. 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](http://www.nsgc.org) and click on “Find a Genetic Counselor.”

A geneticist may help with the initial diagnosis, medical assessment, and coordination of appropriate referrals based on what is known about a child’s medical concerns related to the diagnosis.

**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 a 16p11.2 duplication may struggle with handwriting, using scissors, turning pages, or using a computer. An occupational therapist can work with your child to improve their skills in areas like these.

**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 and coordination.
**Nutritionists** can help children manage their weight. Having 16p11.2 duplication syndrome does not mean the person will be underweight, but it may take more work for them to gain weight. A nutritionist can help a family develop the right eating and nutrition plan for the child with a 16p11.2 duplication.

**Developmental pediatricians** are physicians who specialize in developmental and behavioral disorders in children, including autism, developmental delay, and intellectual disability. They can recommend 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 neuropsychologists** can assess, diagnose and treat a wide range of cognitive, learning, emotional, and behavioral problems.

**Social workers** help families to coordinate care and can provide supportive counseling.

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

**Applied behavior analysis (ABA) psychologists** focus 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 academic, social, communication, motor, and adaptive skills. Research shows that ABA is helpful for children with autism, and for teaching language and other skills to children with developmental delay/intellectual disability.

**Speech-language pathologists (SLPs)** Many people with 16p11.2 duplication syndrome work with a SLP. SLPs help children and adults with a variety of communication, reading, and swallowing problems. The speech and language diagnoses that are common among children with a 16p11.2 duplication are listed below (from the American Speech Language Hearing Association).

**Speech Disorders**

- Speech sound disorders (articulation)
- Control of the muscles of the face and mouth (orofacial myofunctional disorders, dysarthria)
- Difficulty planning and coordinating the movements needed to make speech sounds (apraxia)
- Stuttering
- Voice disorders
Language Disorders

- Difficulty understanding language (receptive language)
- Difficulty using language (expressive language)
- Social communication disorder (pragmatic language)

Other Disorders Treated by Speech-Language Pathologists

- Deafness and hearing loss. Therapy includes developing lip-reading and speech skills, and/or using alternative communication systems
- Oral-motor disorders: weak tongue and/or lip muscles
- Swallowing/feeding disorders

If you are concerned about your child’s communication skills, you may wish to consult an SLP. Your school or school district should have a certified SLP who 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 the department of hearing and speech, clinic for communication disorders, or developmental clinic. Your child’s primary care provider can also recommend an SLP. A search engine from the American Speech-Language-Hearing Association can help you find a certified SLP in your area: http://www.asha.org/findpro/.

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 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. While not all children with 16p11.2 duplication have autism, most will have developmental delay or intellectual disability and will need an IEP at school. 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 psychoeducational assessment and tests to identify strengths and weaknesses is a good starting point. If you would like your child’s school to complete this assessment, you must submit your request in writing. The school should monitor your child’s progress, and your child should receive an IEP and/or curricular modifications. Outside school, many children benefit from additional help from a tutor or aide to encourage and reinforce skills.

Some children participate in a general education classroom and may receive supplemental learning 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.

Some children with 16p11.2 duplication syndrome may not have significant learning problems but may still benefit from receiving accommodations such as receiving extra time on tests or taking tests in a quiet or private space. These children often receive these services through a 504 plan. The school system can provide a 504 plan.
Adaptive Technologies

There are numerous technologies that can help a child with 16p11.2 duplication syndrome with learning and language skills. To find out what adaptive technologies may help your child most, request an assistive technology (AT) or augmentative and alternative communication (AAC) evaluation from a skilled clinician.

When selecting a speech device, take the following points into account:

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

(Information adapted from Assistive Technology: A Parent’s Guide.)

Learn more about these devices here.

Tablets and iPads can now be used in assistive technology. While Simons Searchlight researchers and clinicians do not endorse any specific assistive devices, you may wish to watch our webinar on AAC and on selecting an iPad.

In this webinar, Kelly A. Johnson, Ph.D., of the University of Washington Autism Center discusses how to choose a tablet and applications (apps) for your child to use as a communication device. For example, it’s important to consider what size tablet will best fit your child’s needs. For example, a child with poor hearing will benefit from having a larger device with larger speakers. The amount of storage needed, built-in features, accessories, 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 them to get the full benefit you will both need expert guidance on which apps to use to reach goals. Apps can be used for communication, social interaction, education, and even distraction during medical procedures. When choosing apps for your child to use, bear in mind that the ones that cost money are not necessarily the most useful. While many apps cater 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 connection. If a child is using an iPad for communication, no one other than the child should use it. Also bear in mind that the iPad is not the only useful technology out there.

Learn more about the latest and most liked apps at:

- Mac/Apple http://www.apple.com/education/special-education/ios/
Acknowledgments

There are many people to thank in a project as large as this. First and foremost, we want to acknowledge the sponsor of Simons Searchlight, the Simons Foundation Autism Research Initiative. Its commitment to improving the understanding, diagnosis, and treatment of autism 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 Searchlight study team — including the investigators, study coordinators, project managers, and administrators, as well as our website developer, Tempus Dynamics — who have dedicated many hours to meeting with families and making sure the project runs smoothly. Their work, along with that of other collaborators, has led to numerous publications about 16p11.2 CNVs (listed below).

Most importantly, we owe a huge debt of gratitude to the many families who have participated in the Simons Searchlight study. Since 2010, over 160 people with a 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. Thank you for your contribution, Elly. Your work has had a tremendous impact.
## Resources

### Web resources

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<tr>
<td>The Simons Searchlight website</td>
<td><a href="http://www.simonssearchlight.org/">www.simonssearchlight.org/</a></td>
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<tr>
<td>The Simons Searchlight (and Simons VIP Connect) webinars archive</td>
<td><a href="http://www.youtube.com/c/simonssearchlight">www.youtube.com/c/simonssearchlight</a></td>
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<td>Unique – Understanding Chromosome Disorders</td>
<td><a href="http://www.rarechromo.org">www.rarechromo.org</a></td>
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<td>Genetic and Rare Diseases Information Center – 16p11.2 Duplication</td>
<td><a href="http://www.rarediseases.info.nih.gov/gard/12388/16p112-duplication/resources/1">www.rarediseases.info.nih.gov/gard/12388/16p112-duplication/resources/1</a></td>
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## References

### Articles Using Simons Searchlight Data

<table>
<thead>
<tr>
<th>Year Published</th>
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<tbody>
<tr>
<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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<tr>
<td>2014</td>
<td>Opposing brain differences in 16p11.2 deletion and duplication carriers</td>
<td>Qureshi A.Y. et al.</td>
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<td><a href="http://www.ncbi.nlm.nih.gov/pmc/articles/PMC4138332/">www.ncbi.nlm.nih.gov/pmc/articles/PMC4138332/</a></td>
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<td>Reciprocal white matter alterations due to 16p11.2 chromosomal deletions versus duplications</td>
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<td>A potential contributory role for ciliary dysfunction in the 16p11.2 600 kb BP4-BP5 pathology</td>
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<td>Modulation of mu attenuation to social stimuli in children and adults with 16p11.2 deletions and duplications</td>
<td>Hudac et al.</td>
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<td>2016</td>
<td>Autism spectrum disorder, developmental and psychiatric features in 16p11.2 duplication</td>
<td>Snyder et al.</td>
</tr>
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<td>2019</td>
<td>Psychotic symptoms in 16p11.2 copy number variant carriers</td>
<td>Jutla et al.</td>
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<td>2019</td>
<td>Atypical neural variability in carriers of 16p11.2 copy number variants</td>
<td>Al-Jawahiri et al.</td>
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Other Articles About 16p11.2 Duplication Syndrome

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<th>Year Published</th>
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<tr>
<td>2008</td>
<td>Association between microdeletion and microduplication at 16p11.2 and autism</td>
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<td>2009</td>
<td>Microduplications of 16p11. 2 are associated with schizophrenia</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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<tr>
<td>Year</td>
<td>Study Title</td>
<td>Authors</td>
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<tr>
<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>Rare and common variants at 16p11.2 are associated with schizophrenia</td>
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