

MECP2 Duplication *at a Glance*

Note: These pages are for MECP2 Duplication. If you are looking for Rett/MECP2 Related Disorders, please click [here](#)

MECP2 duplication is a complex neurodevelopmental genetic condition. It is characterized by low muscle tone as infants, moderate to severe intellectual disability, difficulty developing speech, muscle spasticity that is progressive, recurring respiratory infections, and seizures. This condition is caused by a change on the X chromosome. MECP2 duplication syndrome is expressed almost exclusively in males, however rarely girls with MECP2 duplications can show some of the features.

MECP2 duplication syndrome was first described in 2005 and studies now suggest that 1% of cases of X-linked intellectual disability may be due to this syndrome.

Learn more about the clinical features of MECP2 duplication

Common features of MECP2 duplication syndrome in boys (NOTE: not all boys with MECP2 will have all of these features and only rarely will girls have any of these features)

- Significant intellectual disability (100%) with limited or absent speech.
 - Some may experience developmental regression
- Early onset hypotonia (low muscle tone) with slow motor development
 - Sitting, crawling, and walking are very delayed or challenging
 - 2/3 of boys may be able to walk
- Progressive spasticity of the lower limbs (usually more in legs than arms)
 - Ataxia (uncoordinated movement - particularly with walking)
 - 75% get infections easily and often
 - Most likely occur as recurrent respiratory infections
- Seizures occur in 50%
- Autistic-like behaviors may be present
 - Repetitive movements of hands
 - Teeth grinding
- Gastrointestinal dysfunction may be present
 - Constipation
 - Reflux
 - Bladder issues

- Mild facial differences
 - Flat head
 - Underdevelopment of the middle of the face
 - Large ears
 - Flat nasal bridge

Things to Think About

1. Medical and Dietary Needs

What You Need to Know

The list of possible medical problems in MECP2 duplication can be quite extensive. However, each individual usually has only some of these problems. Also, the severity of any one of these medical problems varies widely. Therefore, it is important to ask the parents about the medical issues for their child.

School age boys who have MECP2 duplication should have annual doctor and, often, specialist's visits to monitor medical conditions.

Many boys (about 50%) with MECP2 duplication will have seizures. The seizures are most often tonic-clonic seizures. The children are very likely to be on drugs that control seizures but the seizures may not respond well to the drugs. Typically, the earlier the onset and more severe the seizures, the greater the impact is on the nervous system.

Recurrent respiratory infections, especially recurrent pneumonia, occur in about 75% of boys. These infections may require assisted ventilation and may be fatal. It is important to intervene early if a respiratory infection occurs.

Hypotonia may have already progressed to spasticity by the time boys with MECP2 duplication are school-aged. This can lead to uncoordinated movement (ataxia) especially in the lower limbs. While 2/3 of boys will learn to walk, they may progress to a wheelchair by adulthood. The remaining third are never ambulatory.

What you can do

- Report any change in seizure activity to the parents. Follow school protocols when seizures do occur.
- Ensure a yearly check up in the child's medical home.

- Ensure up to date immunizations. Most children with MECP2 duplication can receive live virus vaccinations. Record types of vaccinations the child receives.
- Support good hand washing to reduce the spread of viruses.
- Notify parents of changes in energy level.
- Be aware of any changes in behavior or mood. Notify the parents.
- Be aware of any changes in academic performance. Contact parents.
- Be an advocate for the child who uses communication supports so that the child can communicate effectively throughout the day. This may include alternative and augmentative communication systems or devices.
- Dietary: GERD (gastroesophageal reflux) may occur. Talk with the parents about particular foods that might be triggers for the reflux and avoid those foods. If the child has more vomiting or reflux than normal, contact the parents so that the cause can be determined.
- Physical Therapy: Stretching exercises can help maintain joint range of motion, prevent secondary contractures, and prolong ability to walk
- Physical accommodations for boys who are non-ambulatory may be necessary

2. Education Supports

It is important to have HIGH LEARNING EXPECTATIONS for children who have MECP2. Encourage use of the core educational curriculum and modify it in order to meet the individual needs of the child.

What you need to know

Individualized, flexible, and appropriate educational strategies/supports are keys to success

- Intellectual ability may be underestimated due to lower functional abilities.
 - Developmental testing may be difficult because of attention, activity, speech and motor issues.
 - Formalized testing has limitations. Make sure testing includes observing the child's performance.

Speech

- Speech development is very delayed and the majority of children do not develop speech
 - Some boys who speak with a few words in childhood may progressively lose speech during adolescence.
 - A small number of children will learn to use 1 or 2 words consistently

- May communicate by pointing, using gestures, and by using communication boards and AAC devices.
- When children have difficulty communicating, they may resort to behaviors such as pulling hair, hitting, biting.
 - Make sure they have a communication system that is effective for their needs.
 - Frustration with communication is often the reason for negative behavior.
 - May not need a behavior plan but rather an effective communication system.

Intelligence

- Most boys will have significant intellectual disability (moderate to severe)
 - Girls with MECP2 duplication may have cognitive challenges of varying degrees, with or without seizures.

Attention

- They may pay more attention when they are naturally curious
- High interest in communication is a sign that child is ready to learn sign language and other ways to communicate.

What You Can Do

Speech and Communication

- Teach learning strategies for non-verbal expression.
 - Consider new technology, computers, and possibly sign language depending on fine motor skills. Focus on *non-verbal* methods of communication.
 - Use augmentative communication aids such as picture cards or communication boards early.
 - Communication should work with child's desire to socially interact with others in natural settings.
 - Make sure they have opportunities for choice and control in their lives (choose books, colors, clothing, play, work partner, etc.)
 - Need multiple means of communication paired with the knowledge of when to use one method vs. another.
 - Find AAC system that allows for maximal social reciprocal communication.
 - Encourage finger pointing early to help with device use as they age.
 - Anyone interacting with the child should have education and training on how to encourage reciprocal communication with the device.
 - Model, model, model the use of the AAC device to encourage its use.

- Continue with strategies that improve oral control to maximize their potential as oral speakers.

Movement

- Unstable or non-walking children may benefit from physical supports in the classroom.
- They may need extra supports/people to help them in their academic program and to be fully included.
- Children with more motor issues may need extra space and/or minimal obstructions to be safe.
- Physical, speech, and occupational therapy to enable walking, proper positioning, hand use, communication needs, etc.
- Bracing and surgery may be needed to align legs.
- Ensure all areas are safe, free of obstacles.

3. Behavioral and Sensory Supports

Individuals with MECP2 duplication should have neuropsychological evaluation to assess abilities and offer support for behavior challenges. Boys with this duplication may have autistic-like features (including anxiety and stereotypic hand movements). Female carriers of MECP2 duplication who have normal intelligence may have neuropsychiatric symptoms including depression, anxiety, and autistic features.

What you need to know

Social

- Social opportunities allow children to express a broad range of feelings and form close bonds and real friendships with others.
- They should be part of typical family and class activities, household chores, and perform daily living skills.
- They may have interests in recreation, music, and physical activity.
- Behavioral supports may be helpful in limiting the less desirable behaviors that are socially disruptive and/or self-injurious.

What you can do

- Be proactive with behavioral supports.
 - Discuss involvement of behavioral or mental health professionals, and/or medications with the parents as needed.
 - Firm directions, rules, and clear expectations are helpful.

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- Many children have difficulty regulating emotions and behavior. This is especially true when handling unplanned changes.
 - Talk through expected changes.
 - They usually thrive with consistency and routine. They can be easily upset with disruption.
 - Prepare for any change in schedule.
 - Provide a safe area to share emotions.
 - Teach and model use of words and/or pictures in sharing emotions.
 - Teach, emphasize, and reinforce behaviors you want to see.
 - Make sure they have an effective communication system.
- Provide social cues and coaching.
 - Provide information to and discuss differences with the child's peers.
 - Help develop confidence and focus on strengths.
 - Provide positive reinforcement.
 - Teach appropriate social behaviors/skills (e.g., how to ask a friend to play).
 - Teach how to recognize facial expressions, body language, and moods in others.
 - Teach how to regulate own body – sensory strategies may be helpful.

4. Physical Activity, Trips, Events

What you need to know

- Any change in routine may produce anxiety, fears, and/or worry. Crowds and loud noise may be hard for some children.

What you can do

- Be proactive and discuss any change in schedule or setting with the child ahead of time.
- Use social stories and pictures to help them understand the change.
- Encourage use of their communication system to help them process concerns.

5. School Absences and Fatigue

What You Need to Know

- Children with MECP2 duplication may be absent due to illness and/or medical appointment
 - Help to make transitions in and out of school as seamless as possible

- Children with MECP2 duplication may be tired and require rest opportunities or breaks in their day

6. Emergency Planning

What You Need to Know

Emergency plans will be individually determined, based on behaviors and medical issues. It is important to mention new signs, symptoms, or pain to the child's parents.

7. Resources

Genetics Home Reference

Consumer-friendly information about human genetics from the U.S. National Library of Medicine

<https://ghr.nlm.nih.gov/condition/mecp2-duplication-syndrome>

GeneReviews

More information on the genetics of MECP2 Duplication Syndrome at Gene Reviews

<http://www.ncbi.nlm.nih.gov/books/NBK1284/>

MECP2 Duplication Syndrome Family Support

The site for families affected by MECP2 <http://www.mecp2duplication.com>

MECP2 Duplication Syndrome Blog

This blog is intended to share information of interest to families affected by MECP2 Duplication Syndrome and others who are concerned about those with this condition.

<http://mecp2.wordpress.com/>