

## MECP2 Duplication *For Healthcare Providers*

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools ([www.gemssforschools.org](http://www.gemssforschools.org)).

The *MECP2* duplication syndrome is a severe neurodevelopmental disorder involving a duplication of a gene on the X chromosome, previously identified with mutations in Rett syndrome in females and often lethal in males. *MECP2* duplication syndrome is **NOT** Rett syndrome. *MECP2* duplication syndrome affects 100% of males and is asymptomatic in almost all females. Most males with *MECP2* duplication syndrome are born to carrier females. That is, mothers who have the same duplication. Almost 50% of males with *MECP2* duplication die before the age of 25 years, either from complications from infections or from neurologic deterioration. With the identification of more affected individuals, phenotypic variability has been noted.

### Physical characteristics and/or symptoms

*Note: not all boys with MECP2 Duplication will have all of these features, and girls will rarely have any of these features.*

#### Core features of MEPC2 duplication syndrome in boys

- Moderate to severe intellectual disability
  - Limited or absent speech.
- Infantile hypotonia with delayed motor development
  - Only 1/3 of males learn to walk
- Progressive spasticity of the lower limbs
- Seizures
  - Generalized tonic-clonic
  - Absence seizures
- Recurrent respiratory infections

#### Other features

- Autistic-like behaviors may be present
  - Repetitive movements of hands
  - Teeth grinding

- Gastrointestinal dysfunction
  - Constipation
  - Reflux
  - Bladder issues
- Mild facial dysmorphism

## Recommended Routine Surveillance

- Monitor for progressive spasticity
- Monitor for seizures
- Monitor gastrointestinal signs
- Monitor for recurrent respiratory infections
- Monitor for development

## Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Emergencies should be handled as with any child.
- If seizures are present, the following seizure action plan may be useful:  
[https://www.aap.org/en-us/Documents/Seizure Action Plan for%20School.pdf](https://www.aap.org/en-us/Documents/Seizure_Action_Plan_for%20School.pdf)

## Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Developmental evaluation:
  - Speech therapy with emphasis on nonverbal methods of communication
  - Physical therapy
  - Occupational therapy
  - Autism spectrum disorder /autistic signs
- Gastroenterologist
  - Feeding difficulties
  - Gastrointestinal reflex
- Geneticist / Genetic Counselor:
  - Diagnosis
  - Coordination of care
  - Genetic risk for family
  - Clinical trials

- Neurologist
  - Monitor seizures
- Pulmonologist
  - Recurrent pulmonary infections

## Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

*My patient \_\_\_\_\_ has been diagnosed with MECP2 duplication. This condition is characterized by hypotonia, progressive spasticity, seizures, autistic like features, developmental delays. Medical complications with MECP2 duplication include management of seizures, recurrent infections, and gastrointestinal symptoms. Because of these, \_\_\_\_\_ needs the following accommodations.*

## Seven Important Aspects of School Life

“[MECP2 Duplication at a Glance](#)” will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions



## Resources

### Gene Reviews

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