MOEBIUS SYNDROME FOUNDATION

A GUIDE FOR HEALTHCARE PROVIDERS

This booklet is designed to help healthcare providers get a brief overview of Moebius syndrome and guidance on the types of specialist care a patient may require.

The Moebius Syndrome Foundation gratefully acknowledges the contributions of the following members of our Scientific Advisory Board.

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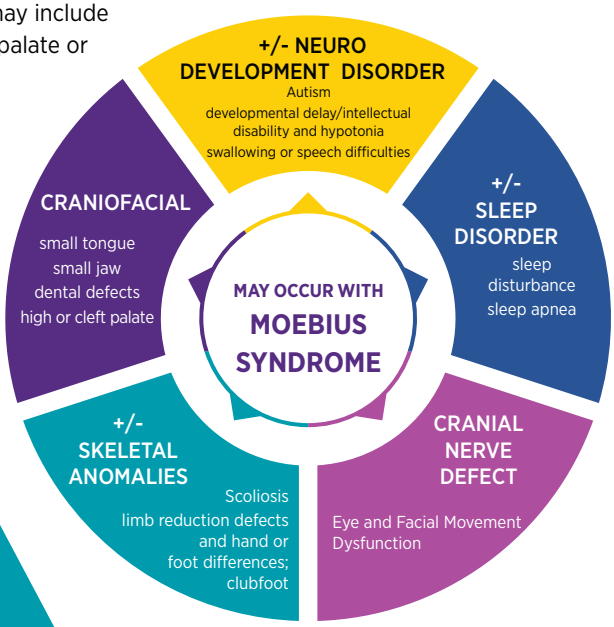
WHAT IS MOEBIUS SYNDROME?

Moebius syndrome is a rare, congenital, non-progressive neurological disorder characterized by facial paralysis and abnormal ocular motor function. Clinically, individuals with Moebius cannot smile, frown, blink, or move their eyes outward to the sides. Most commonly, the disorder is bilateral, creating an expressionless appearance. In the newborn, this disorder may be suspected due to a lack of facial grimace when crying, poor sucking or an inability to nurse, and ocular misalignment.

In the classic form, Moebius syndrome is due to the paralysis of the motor neurons of the sixth and seventh cranial nerves. The abducens nerve (CN VI) controls the eye muscles which move the eyes outward. The facial nerve (CN VII) controls the muscles to the forehead, eyelids, cheeks, and lips.

More complex cases of Moebius may include facial abnormalities, such as cleft palate or a small tongue and jaw, which may cause upper airway respiratory compromise. Poland syndrome or additional cranial nerve involvement may be seen. Limb differences may occur; clubfoot is the most common lower limb problem. Upper limb differences may include web fingers or missing digits.

First described in 1880, the exact anatomical nature of the neurological defect causing Moebius syndrome remains poorly defined. Incomplete development or calcifications of the involved cranial nerves or the associated cranial nerve nuclei have been reported. Cases are sporadic and seen equally in male and females, and in all ethnic groups. The cause is unknown, although vascular disruption of embryogenesis in weeks 7-12 is postulated.

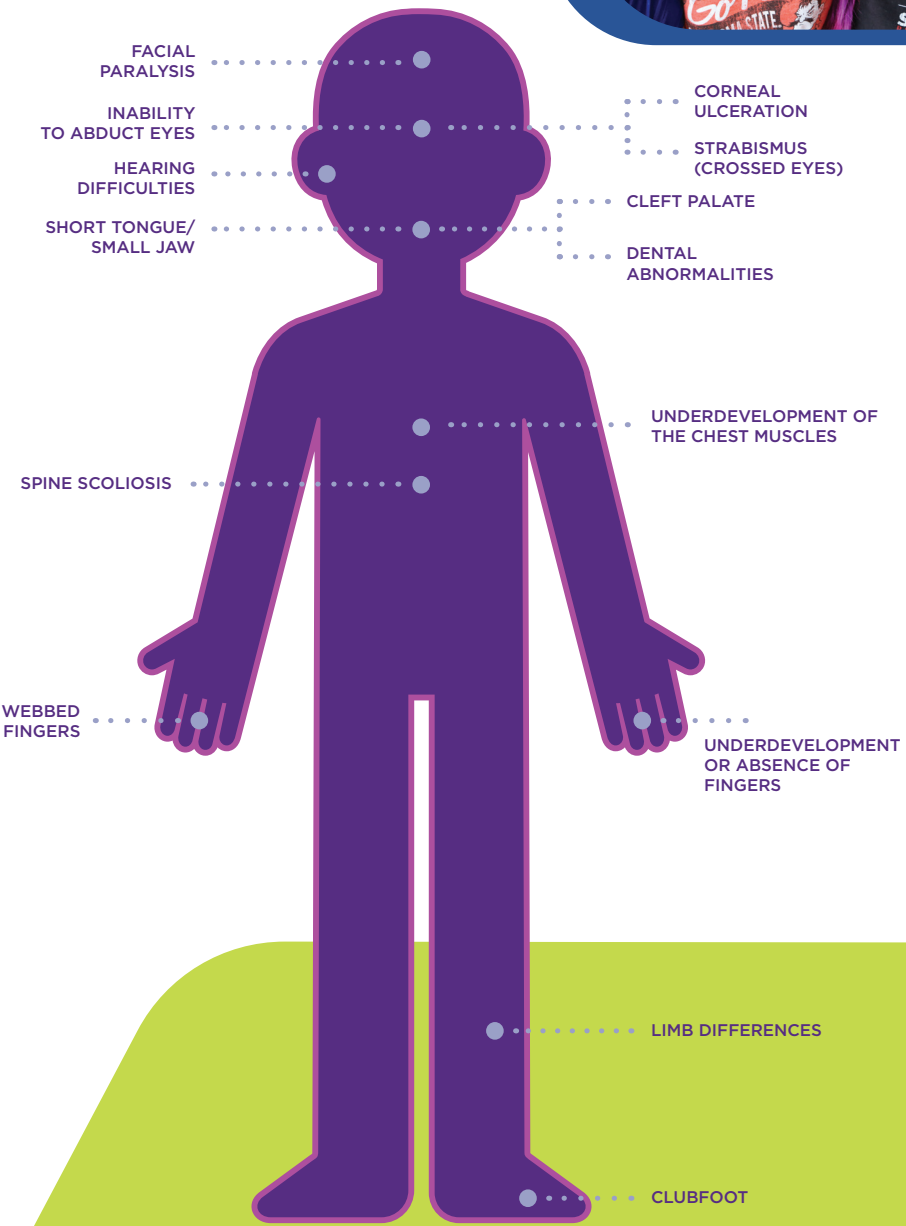


SCAN TO LEARN MORE ABOUT MSF

MOEBIUS SYNDROME FOUNDATION

Does not include all symptoms that occur with Moebius syndrome.

COMMON PHYSICAL SYMPTOMS



HOW TO GET A DIAGNOSIS

In 2007, scientific researchers at the Moebius Syndrome Foundation Research Conference defined a diagnosis of Moebius syndrome as twofold:

1. Congenital facial weakness (present at birth)
2. The inability to abduct (move the eye away from the nose) one or both eyes

A diagnosis for Moebius syndrome can be made by any physician (ex., geneticist, neurologist, etc.) as long as the clinical definition is met.

Neurology helps us understand the structure of the cranial nerves and the brain. A neurologist may order an MRI to evaluate the 6th and 7th cranial nerves. An MRI may help determine if the nerves are fully developed and intact or are underdeveloped or missing.

Ophthalmology will examine the eyes and how they move. An ophthalmologist can determine if a patient has the inability to abduct (move the eye away from the nose) one or both eyes.

Genetic conditions may also need to be ruled out. Some genetic disorders may present like Moebius syndrome, e.g. *TUBB3* variants. A geneticist may take a saliva sample or draw blood to study the genetic makeup of a patient and help shape a Moebius syndrome diagnosis. There is no known genetic marker for Moebius itself at this time.

These two symptoms may be due to the underdevelopment or absence of the facial nerve (cranial nerve 7) and the abducens nerve (cranial nerve 6), respectively. There is a fairly long list of co-symptoms with Moebius and it presents differently in every individual. Club feet, limb abnormalities, and hypotonia are all very common with Moebius syndrome.

SCAN FOR GENETIC RESEARCH UPDATE

EYE CONCERNS

Considering that an abduction deficit (limitation of outward eye movement due to 6th nerve palsy) is one of the diagnostic criteria for Moebius syndrome, virtually every patient with Moebius has a profound limitation of abduction. Despite this limitation, up to 30% may have straight eyes. The remainder have strabismus, typically esotropia (crossed eyes). Esotropia may interfere with social interactions, reduces the functional visual field, and reduces stereopsis (depth perception) contributing to gross motor delays. Surgery is therefore offered to patients to reduce the estropia (though it does not restore normal eye movement). The medial rectus muscle can be very tight in these cases, making surgery technically challenging and increasing the complication rate. In more severe cases, transposition of the extraocular muscles provides a small amount of abduction and stabilizes eye alignment over time. These technical challenges make it important to have an experienced strabismus surgeon perform the operation.

The other common eye issue in patients with Moebius is dry eyes. This dryness is due both to incomplete or ineffective blinking during the day and to corneal exposure while sleeping. The severity of dryness ranges from mild irritation to vision-threatening corneal ulcers requiring management of a specialist in corneal diseases and use of specialty contact lenses. Even patients with minimal or no symptoms should consider using a lubricating gel at night to prevent long-term complications.

THE GOOD NEWS IS THAT DESPITE THESE CHALLENGES, VISUAL FUNCTION IN MOST PATIENTS WITH MOEBIUS IS GENERALLY GOOD ENOUGH TO ALLOW FOR READING, DRIVING, AND OTHER VISUAL ACTIVITIES.

SCAN TO LEARN MORE ABOUT EYE BASICS FOR PEOPLE WITH MOEBIUS SYNDROME

SPEECH AND FEEDING

The effects of Moebius on feeding and speech varies from person to person. For some, lack of upper lip movement affecting mouth closure is the primary area of difficulty. With others, jaw control and tongue control may be impacted as well.

The overall health of the oral cavity is important. A healthy oral cavity starts with the ability to maintain lip closure and the tongue against the palate, which allows nasal breathing.

Working with a speech and language pathologist that understands motor development can positively impact families and individuals with Moebius. The more families develop their understanding of how motor skills develop, the better informed they are on how they can help their children potentially develop these skills through additional therapy.

Many people with Moebius have difficulty with motor skills from infancy. Motor development delays can lead to further problems as they develop in speech clarity, oral rest posture, and eating skills. Oral motor skills develop through early feeding, which is often impacted by having Moebius syndrome. When there are feeding problems, which may impact nutrition and swallowing safety or oral development, it can lead to further problems in speech clarity. Although muscles can change at any age, starting early and working preventatively is best.

TRADITIONAL SPEECH AND FEEDING TECHNIQUES OFTEN NEED TO BE ADAPTED AND IMPLEMENTED USING A SENSORY MOTOR APPROACH TO GAIN MUSCLE ACTIVATION. INCREASED INPUT TO THE SENSORY SYSTEM CAN BE KEY TO GAINING MOTOR MOVEMENT.

SCAN FOR SPEECH AND FEEDING TECHNIQUES

SMILE & BLINK SURGERY

Patients with Moebius syndrome may benefit from reconstructive surgery to provide smiling movements. For many years, this surgery has been accomplished by introducing new muscles into the face with muscle transplants. Often, a small segment of the gracilis muscle, a small muscle in the inner thigh, is used. One of the most common methods to produce movement, is a nerve that is ordinarily used for chewing, is frequently repurposed to activate the gracilis muscle transplant. Therefore, patients initially need to bite down to smile, and, with training and time, adapt to smiling without needing to bite down. These operations are long lasting and only use muscles and nerves from the patient's own body. Each operation takes about six hours to perform, and patients generally stay in the hospital three or four days. It usually takes a couple of months for new smiling movements to begin, and then a gentle exercise program is used. The second side is generally treated about three months after the first.

More recently, a new blinking surgery has been developed to enable greater eyelid closure. Blinking is important because it protects the eye surface. Blinking also facilitates communication with other people. This operation involves nerve transplants taken from the lower leg during surgery. The nerves are connected to a different chewing nerve and the nerve cells grow into the eyelid regions. Then a muscle transplant is performed months later. The nerve impulses enable the muscles to contract and facilitates eyelid movements. These operations can be done at the same time as smile surgery and can also be done for patients that have previously undergone smile surgery.

COMMON DENTAL CONCERNS

Many individuals with Moebius syndrome have difficulty in closing their mouth or swallowing. Microstomia is common. The tongue may be larger or smaller than average or unusually shaped. The tongue is often hypotonic or it may fasciculate. The palate may be narrow and arched excessively since the tongue does not form a suction that would usually shape the palate more favorably. Cleft palate is a possibility.

Micrognathia or retrognathia are often seen. There is a tendency for an anterior open bite. Misalignment of teeth may develop. Interceptive orthodontic treatment is helpful. Sometimes orthognathic surgery is necessary.

The saliva may be thick, or a dry mouth may be present. Excessive drooling can be a problem too. Food may remain in the pouches and sides of the cheek after eating. This needs to be cleared with a toothbrush or gloved finger and not allowed to remain in the mouth. There may be enamel hypoplasia. All of these factors increase the vulnerability to dental caries. Reduce sugarladen foods and snacks to prevent additional wearing.

THE FIRST APPOINTMENT WITH A DENTIST SHOULD BE AS EARLY AS ONE YEAR OF AGE.

A dentist may use a rubber dam to prevent aspiration and help control tongue interference with treatment. Atraumatic RestorativeTreatment, including Silver Diamine Fluoride, could be beneficial. Sedation may be needed. Dental implants can help with replacing teeth.

Each person with Moebius Syndrome is unique and everyone's dental concerns are best addressed in consultation with a dentist who has evaluated the patient.

SCAN FOR ORAL HYGEINE TIPS

RESPIRATORY & PULMONARY CONCERNS

Patients with Moebius syndrome have many reasons for getting respiratory symptoms, and they are not always recognized as a problem or something that can be helped.

Some patients are at risk for aspiration with drinking and with gastroesophageal reflux. It is worth asking them if there is any coughing with drinking and whether they have symptoms of reflux. Some have long-standing reflux and will complain that their food "gets stuck" after they swallow.

Because of chronic aspiration, many patients with Moebius develop reactive airway disease. They should be asked about whether they have shortness of breath with exercise or related to the weather. Also, patients may cough excessively with colds and during allergy season. Some patients with Moebius have excessive mucus in their lungs, may choke on it, and may be prone to pneumonia.

Also related to Moebius syndrome, patients can have either central or obstructive sleep apnea. They should be asked about being restless during sleep, having difficulties waking up in the morning, having morning headaches, and also having daytime sleepiness. They may need a sleep study.

IT IS IMPORTANT TO REMEMBER THAT PATIENTS WITH MOEBIUS SYNDROME ARE AT HIGHER RISK THAN THE GENERAL POPULATION FOR ASPIRATION, RECURRENT PNEUMONIA, AND SLEEP APNEA.

Because of narrowed airways and difficulty swallowing, tracheostomy (trachs) and feeding tubes may be necessary in patients with Moebius syndrome. Also, it should be noted that intubation, either for surgery or for respiratory support, may be very difficult. Direct visualization may be needed. Anesthesia services need to be familiar with the patient's diagnosis and special needs prior to any procedure that requires anesthesia or sedation.

SCAN FOR MANAGEMENT OF RESPIRATORY SYMPTOMS

ORTHOPEDIC & PHYSICAL THERAPIES

The musculoskeletal manifestations of Moebius syndrome vary widely between individuals. An increased rate of clubfoot (a stiff foot in a turned-in position), scoliosis (curvature of the spine), Poland syndrome (chest muscle weakness with differences in hand anatomy), transverse limb deficiencies (congenital amputation or missing digits), and hip dysplasia have been reported. An orthopedic surgeon experienced with these differences may need to be involved in the care of people with Moebius.

Clubfoot in patients with Moebius syndrome has an increased chance of needing surgical care, but typically responds to Ponseti casting initially. Bracing the feet after correction is of paramount importance to prevent recurrence and minimize the need for surgical treatment. Scoliosis (lateral curvature of the spine) is generally noticed in the early teenage years, and, in most instances, may be managed with a brace. In some cases, surgical treatment is required.

Poland syndrome (weakness in the chest muscle and hand differences) may require the attention of a surgeon as well, and varies broadly among patients with Moebius syndrome.

GENERALIZED "LOOSE JOINTS" ARE NOT UNCOMMON – PHYSICAL THERAPY AND STRENGTH TRAINING CAN BE HELPFUL IN ALLEVIATING SYMPTOMS THAT RESULT.

Central to any surgical treatment is proper anesthetic care. Patients with Moebius syndrome may have smaller mouths and jaws than average, which can lead to difficulty with placing a breathing tube during surgery. It is important to consult with your surgeon and anesthesiologist to be sure that adequate equipment and expertise are available.

SCAN FOR OUR WEBINAR WITH DR. PHILIP K. MCCLURE

MENTAL HEALTH

MENTORSHIP CAN BE REQUESTED AT BIT.LY/MSFMENTOR

The majority of people with Moebius syndrome live well. However, Moebius syndrome (MBS) may increase the risk for mental or psychological health problems, including:

- Anxiety and depression
- Sleep difficulties
- Social problems, such as stigmatization, staring, or bullying
- Neurocognitive disorders, such as a learning disorder (LD)
- Some research suggests a higher rate of autism spectrum disorder (ASD), although care must be taken during an assessment to ensure that symptoms of MBS (facial paralysis, limited eye movement, and speech delay) are not mistaken for autism symptoms.

By informing patients of the mental health risks, practitioners are able to monitor the patient and refer professional assessment and intervention as indicated. Early intervention is the best possible course of action, so caregivers should monitor closely for developmental differences or delayed learning. Medical team members, Early Childhood Intervention (ECI) professionals, and educators are available to assess and guide parents accordingly. Social and emotional problems are not present at birth but rather develop over time, as living with differences in physical appearance and functional abilities can be stressful. And across time, a high level of stress may lead to social and emotional problems. Skills to compensate or cope with these stressors can decrease or eliminate social and emotional problems. Humans use a vast array of communication channels, including gesture, prosody (tone of voice), language, posture, proximity, style, and facial expression. Many people with Moebius naturally increase their expression through channels other than the face or can learn to do so through social skills training. This is an effective strategy; research shows that people with Moebius who use this type of expression are rated more positively by strangers than those who use less.

Mental health professionals and Moebius community mentors are able to teach compensatory and coping skills for living with differences in physical appearance and functional abilities. Medical team members are able to provide referrals to mental health professionals.

CHECK OUT THIS ARTICLE BY ADVISORY BOARD MEMBER, KATHLEEN R. BOGART PH.D

Disclosing the Obvious:
Explaining Facial Differences

MOEBIUS SYNDROME FOUNDATION

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To learn more about Moebius syndrome, how to collaborate, attend a future Scientific Research Symposium, or about the support and resources the Moebius Syndrome Foundation offers, please contact us:

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Stay updated with our latest news and events. Email newsletter@moebiussyndrome.org to subscribe to our monthly newsletter.

Pictured: Moebius Syndrome Foundation Scientific Advisory Board and research supporters.
The Moebius Syndrome Foundation has invested in ongoing research studies, including research into the cause of Moebius syndrome with a collaboration between the NIH, Boston Children's Hospital and Icahn School of Medicine at Mount Sinai, NY.

Special recognition goes to Rick Guidotti of Positive Exposure for some of the photography contributions.

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