

# MEIER-GORLIN SYNDROME

## DESCRIPTION :

Meier-Gorlin syndrome (MGS), a rare genetic condition, is one of six diagnoses in the "primordial dwarfism" family. This syndrome is subdivided into 8 types, which affect different genes :

- Type 1 affects the *ORC1* gene;
- Type 2, affects the *ORC4* gene;
- Type 3, affects the *ORC6* gene;
- Type 4, affects the *CDT1* gene;
- Type 5, affects the *CDC6* gene;
- Type 6, affects the *GMNN* gene;
- Type 7, affects the *CDC45L* gene;
- Type 8, affects the *MCM5* gene.

In adulthood, the average height of men is 147 cm (4 ft 10 in) and that of women is 138 cm (4 ft 6 in).

All the forms of this syndrome are autosomal recessive traits, except type 6, which is an autosomal dominant trait [refer to the "genetics" factsheet].

The prevalence of the condition is unknown. Currently, around 67 (as of 2015) cases have been reported.

## CHARACTERISTICS AND DIAGNOSIS :

Meier-Gorlin syndrome causes proportionate dwarfism characterized by facial dysmorphism (irregularities of the face). The main features of the diagnosis are as follows:

- Microcephaly (small head);
- Microstomy (small mouth) and full lips;
- Microtia (underdevelopment of the ears);
- Hypoplasia (underdevelopment) of the jaw;
- Palatal fissure (opening of the palate);
- Small appearance and narrow torso (with mammary hypoplasia in women);
- Patellae absent or underdeveloped.

Various skeletal abnormalities are also possible such as dislocation of the elbows, long and slender ribs and other bones, flattening of the epiphyses (head) of long bones, clavicle and finger irregularities.

At birth, most newborns have low birth weight and microtia (underdeveloped ears). The diagnosis is made on the basis of a clinical and radiological examination, as well as an



ultrasound of the kneecaps. The association of microtia/anomaly of the kneecaps/growth impedance is the first criteria to establish the diagnosis of Meier-Gorlin syndrome. A genetic test can confirm the diagnosis.

### MAIN POSSIBLE COMPLICATIONS :

**Jawbone:** If the jawbone is irregular, maxillofacial surgery may be used to correct the issue. Surgery can be performed when the jaw is fully grown.

**Palatal fissure** (opening of the palate): This characteristic feature can cause difficulty swallowing and speaking. It can also cause middle ear dysfunction, which is why the ears and eardrums of infants with this issue should be monitored from the age of nine or 12 months and then every six months thereafter.

Surgery is available to correct the cleft palate.

**Microtia** (underdevelopment of the ear) or **Anotia** (complete absence of the ear): The underdevelopment of the ear can be variable, which is why an audiological assessment, associated with imaging, must be carried out to detect the extent of the damage to the external auditory canal, the middle ear and the inner ear.

Microtia can be corrected with surgical reconstruction of the pinna. This can be done from the age of five, or earlier if needed.

Anotia, the most severe form of microtia, causes deafness, attention deficit disorder, and delayed language development. In this case, wearing a hearing aid is necessary.

**Breast hypoplasia** (in women): Underdevelopment or absence of breasts is common in women with Meier-Gorlin syndrome. Breast augmentation, paid for by Provincial Health Insurance (RAMQ, OHIP etc.) may be considered to remedy this issue.

**Knees:** Underdevelopment or lack of kneecaps can lead to knee instability, pain and early osteoarthritis. It is advisable to have the knees regularly monitored by an orthopedist or a doctor specializing in rehabilitation, and to practice strengthening exercises. The person with MGS must also be careful when participating in sports.

**Feeding Issues:** Children with Meier-Gorlin syndrome have early feeding Issues which can cause serious complications. Pediatricians must closely monitor their patient's diet and growth. Early intervention is important to ensure the health and development of the child.

**Breathing Issues:** 30% to 79% of people with this syndrome have potentially serious breathing difficulties, such as laryngotracheomalacia and pulmonary emphysema. Because of these risks, it is strongly recommended that their respiratory function be regularly assessed, starting at birth. It is also strongly recommended that they be vaccinated against influenza and pneumonia.



- Laryngotracheomalacia is characterized by a softer-than-normal larynx and trachea, this causes airway obstruction and difficulty breathing. This can be a serious issue and can even cause death.
- Pulmonary emphysema is an insidious, incurable degenerative disease that occurs when lung tissue breaks down and loses elasticity. The exhalation then becomes more difficult. Symptoms of this condition include a persistent feeling of shortness of breath, tightness in the chest and fatigue. Breathing can be wheezing in manner, a chronic cough with sputum is present, and the lips may turn a bluish color. Symptoms often appear when the diagnosis is at an advanced stage. The resulting respiratory failure can cause death; it must therefore be followed closely.

**Anesthesia:** The risks of complications for people with Meier-Gorlin syndrome are increased when breathing issues are present.

In the event of a medical operation an experienced anesthetist should be consulted and be present. Finally, before implementing any invasive technique, it is required to have the patient's pulmonary function evaluated [refer to the "anesthesia" factsheet].

#### **TREATMENT :**

The management of Meier-Gorlin syndrome is multidisciplinary (surgery, occupational therapy, orthopedics, pneumology, audiology, physiotherapy, etc.) and preventive. The aim is to detect complications and help those affected to have a better quality of life.

Currently, there is no specific treatment for Meier-Gorlin syndrome. Growth hormone trials are inconclusive. As for limb lengthening, it is not recommended. Indeed, since this form of dwarfism is proportionate, lengthening the legs and arms would create an unbalanced body shape.

**List of the main elements to monitor and address** [refer to the factsheet "Frequent interventions in little people"]:

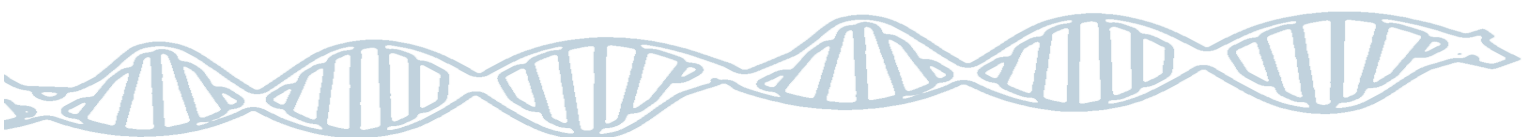
- Palatal fissure;
- Microtia or anotia;
- Feeding problems;
- Breathing difficulties;
- Anesthesia.

#### **RESOURCES :**

Association québécoise des personnes de petite taille

<https://www.aqppt.org/>

Little People of Ontario



<https://littlepeopleofontario.com/>

Regroupement québécois des maladies orphelines - Centre iRARE

<https://rqmo.org/centre-dinformation-et-de-ressources-en-maladies-rares/>

Orphanet

[https://www.orpha.net/consor/cgi-bin/Disease\\_Search.php?](https://www.orpha.net/consor/cgi-bin/Disease_Search.php?)

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NORD - National Organisation for Rare Disorders

<https://rarediseases.org/rare-diseases/ear-patella-short-stature-syndrome/>

GARD – Genetic and Rare Disease Information Center

<https://rarediseases.info.nih.gov/diseases/2033/meier-gorlin-syndrome>

OMIM – Online Mendelian Inheritance in Man

<https://www.omim.org/entry/224690>

Sonja A. de Munnik, Elisabeth H. Hoefsloot, Jolt Roukema, Jeroen Schoots, Nine VAM Knoers, Han G. Brunner, Andrew P. Jackson and Ernie MHF Bongers, “Meier-Gorlin syndrome”, in *Orphanet Journal of Rare Diseases*, 10: 114, 2015

<https://ojrd.biomedcentral.com/articles/10.1186/s13023-015-0322-x>



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Avec la participation financière de

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