

MORQUIO SYNDROME

DESCRIPTION :

Morquio syndrome (also called Mucopolysaccharidosis type 4), a spondylo-epiphyso-metaphyseal dysplasia, is the second most common type of dwarfism in Quebec. The condition, which causes severe dwarfism, is due to mutations in the GALNS gene (in type A of the diagnosis) and the GLB1 gene (in type B of the diagnosis). It results in a defect in the degradation of a mucopolysaccharide (sugar class), also called keratan sulfate.

Growth stops rapidly (at about 8 to 10 years of age), so the average adult height is between 100 cm and 125 cm (3'4" and 4'2").

The condition is an autosomal recessive trait [see Genetics]. It affects approximately 1 in 76,000 to 640,000 births.

CHARACTERISTICS AND DIAGNOSIS :

Morquio Syndrome is a progressive condition that causes disproportionate dwarfism. The physical features of this diagnosis include:

- Limbs of average length, often irregular;
- Short, broad trunk and neck;
- Loose joints (initially);
- Flat feet;
- Hip dislocation;
- Greyish, spotted teeth that decay easily.

Skeletal issues are present and increase as the child grows, including platyspondyly (flattening of the vertebral body of one or more vertebrae), kyphosis (humpback), scoliosis, pectus carinatum (pointed sternum), genu valgum (knees bent inward) and long bone problems.

Morquio syndrome is generally suspected between 1 and 3 years of age due to gait disturbances (waddling gait) and slower growth. The diagnosis is based primarily on urine tests; since undegraded keratan sulfate accumulates in the urine, its unusually high presence is a sign of the condition. The diagnosis is then confirmed by enzyme and genetic tests.

MAIN POSSIBLE COMPLICATIONS :

Eyes: Opacity of the cornea is common. This is not serious and generally poses few problems. There is a risk of photophobia. Surgery is not recommended as recurrence is common. Glaucoma or cataracts may also occur, as well as retinal degeneration.

Eyesight should be evaluated every one or two years. In children over 10 years of age, the evaluation should include tonometry (measurement of eye pressure).



Hearing Issues: Middle ear issues cause progressive hearing loss, which is aggravated by recurrent ear infections. Hearing loss usually occurs between childhood and adolescence and is present in almost all adults over the age of 30.

As a result, a hearing assessment must be performed annually from the time of diagnosis.

Abdomen and hernias: The abdomen is sometimes bulky and swollen. Hepatomegaly (enlargement of the liver) and splenomegaly (enlargement of the spleen) are sometimes present and may cause discomfort and pain.

In addition, children may have one or more hernias. Hernias, located in the groin (inguinal hernia) or navel (umbilical hernia), are particularly visible when the child coughs, cries or does anything to increase intra-abdominal pressure. They can be corrected by surgery, but the risk of recurrence is a consideration.

Respiratory system: Occlusion of the lower and upper airways is common. Affected individuals have laboured breathing and may have hyperextension of the neck.

In particular, breathing issues at night should be monitored, as it is a sign of respiratory failure that can lead to cardiovascular complications. Obstructive sleep apnea is characterized by loud snoring, apneic pauses, compensatory sighs, enuresis ("bed-wetting"), vomiting at night, and irritability or drowsiness during the day [see "Sleep Apnea" fact sheet].

Tonsil and/or adenoid removal, the use of ventilation devices (CPAP or BPBV), or even tracheostomy can be recommended to treat obstructive sleep apnea.

As a preventive measure, pulmonary function tests should be performed in late childhood and repeated every one or two years. In addition, to avoid pulmonary complications related to infections, all individuals should be regularly vaccinated against influenza and pneumococcus.

Heart: Regurgitation or valve blockage, with varying degrees of severity, is common. Valvulopathy can progress slowly over a period of years with no apparent clinical effect. If the problem worsens, it may be necessary to replace the damaged valves through surgery (valvuloplasty). While rare, cardiomyopathy may occur.

The most common symptoms of valvulopathy are discomfort, chest tightness or pressure, palpitations, shortness of breath, fatigue, weakness and dizziness.

Because of the cardiac risks, a cardiological and echocardiographic evaluation should be performed when Morquio Syndrome is diagnosed and preferably every one to two years thereafter.

Neck (cervical spine): This is one of the most serious potential problems of Morquio's disease. From the age of 5 or 6, cervical irregularities can cause instability in the first vertebrae with a risk of compression of the spinal cord. As a result, problems of the spine are common and are often progressive. It is therefore necessary to perform X-rays and MRI (magnetic resonance imaging) tests of the cervical region as soon as the diagnosis of Morquio syndrome is made, and then every year thereafter. In case of acute problems, a surgeon specialized in spinal disorders should be consulted. If necessary, a cervical decompression with fusion may be performed.



Compression of the cervical spine can cause slow and gradual myelopathy, sudden paralysis and even sudden death.

The first signs of myelopathy are decreased muscle endurance and tone, hyperreflexia (exaggerated reflexes) and clonus (rapid contractions and reflexes of the limbs), especially in the legs, and issues with bowel and/or bladder control (incontinence, etc.).

Anesthesia: The risks of complications are numerous and increased due to the cervical, respiratory, cardiac and neurological problems associated with Morquio Syndrome.

In the event of surgery, it is therefore necessary to be sure to call upon the services of an experienced anesthetist [see the "anesthesia" section].

TREATMENT:

Treatment aims to improve the patient's quality of life, slow the progression of the condition and avoid permanent tissue and organ damage. Treatment is therefore mainly symptomatic, focusing in particular on orthopedics (equipment, surgery, neck consolidation by vertebral fusion, etc.) and rehabilitation (physiotherapy, etc.).

In addition, a drug exists to treat type A Morquio Syndrome; VIMIZIM. Unfortunately, this treatment is still not accessible to Quebec patients, except for those who have participated in clinical studies, those who can benefit from reimbursement through their private insurance, or those who have coverage that must be approved under the Patient Exception measure of the Régie de l'assurance maladie du Québec.

List of the main elements to be monitored and addressed [see the "frequent interventions for people of short stature" sheet]:

- Eye and hearing problems;
- Abdominal hernias;
- Respiratory system (especially obstructive sleep apnea);
- Heart;
- Cervical instability with risk of spinal cord compression.

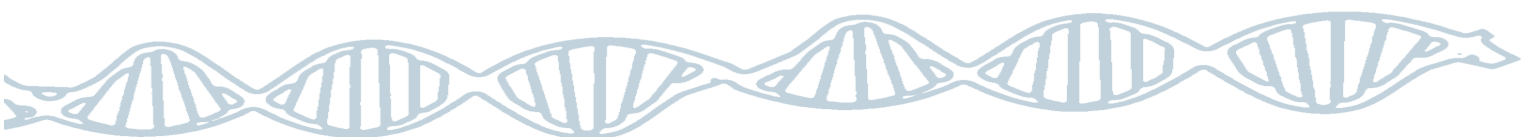
RESOURCES :

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/>

Morquio Community of Quebec

<http://morquioquebec.org/>

Orphanet - Fact sheet on Morquio Syndrome

https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=582&Ing=EN

Little people of America - fact sheet on Morquio Syndrome

<https://www.lpaonline.org/assets/documents/NH%20Morquio%20Syndrome.pdf>

A Guide to Understanding Mucopolysaccharidosis IV, Canadian MPS Society for mucopolysaccharidosis & related diseases, 2013

<http://www.mpsociety.ca/support/resources/mps-iv-resources/>

Living with MPS IV: A Guide for Patients and Families, Canadian MPS Society for Mucopolysaccharidosis & Related Diseases Inc, 2016

<http://www.mpsociety.ca/wp-content/uploads/2018/07/>

MPSIV_binder_20160902_final_FRCA.pdf



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