SECKEL SYNDROME

DESCRIPTION :

Seckel syndrome is one of six diagnoses in the family of "primordial dwarfism". The condition is divided into different types, which involve genetic alterations or mutations in several distinct genes.
Seckel syndrome is caused by osteodysplasia, an issue withbone formation and nutrition. In adulthood, people with Seckel syndrome are often less than 128 cm (4'3") in height.
Extremely rare, the condition affects less than one person in 1,000,000. It is an autosomal recessive trait [see Genetics].

CHARACTERISTICS AND DIAGNOSIS :

Seckel syndrome causes proportionate dwarfism characterized by facial dysmorphia (irregularities of the face). The main features of the diagnosis are as follows:

- Microcephaly (small head);
- Craniosynostosis (premature fusion of one or more cranial sutures);
- Low and receding forehead;
- Proportionately larger nose;
- Retrognathism (posterior positioning of the lower jaw);
- Sharp and misaligned teeth;
- Intellectual disability of varying intensity;
- Sometimes shorter than normal little finger;
- Sometimes clubfoot.

The condition is often discovered at birth because of the baby's facial features, short stature and low weight. However, it is sometimes necessary to wait until the child is older and the full syndrome develops before confirming the diagnosis.

The radiographic findings include, microcephaly (smallness of the head), premature closure of cranial sutures, malformation of the fingers (curvature of the distal phalanges and shortening of the phalanx of the little finger),, dislocation of the head of the forearm on the thumb side (radial dislocation), dislocation of the elbows, dislocation and/or malformation of the hips, and the absence of a pair of ribs (11 instead of 12).

PRIMARY POSSIBLE COMPLICATIONS :

Clubfoot: When clubfoot is detected at birth; it should be managed quickly to avoid complications. Treatment consists of softening and straightening the foot, then applying

adhesive bandages, before putting on casts. At walking age, the child must wear custom-made orthopedic shoes.

Scoliosis or kyphoscoliosis: In some cases, children with Seckel syndrome may develop scoliosis (a three-dimensional spinal problem) or kyphoscoliosis (a double spinal curvature combining scoliosis and a hunched back).
Clinical surveillance must be carried out regularly and must be accompanied by X-rays as soon as any doubt arises.

Eyes: Some people have strabismus (squinting eyes). Eye exercises, glasses, or surgery can correct it if necessary.

Intellectual disability: To some varying degree, intellectual challenges are present in most people with Seckel Syndrome. Often unnoticed at birth, the disability becomes noticeable as the child grows older. Depending on its severity, adapted support may be required.

Hematological abnormalities (blood disorders): Impacts just under 25% of people with Seckel syndrome, but should be taken seriously. The most common diagnoses are:

- Anemia (a decrease in red blood cells): Characterized by pallor, fatigue, headaches, unusual shortness of breath, tachycardia, dizziness and light-headedness.
- Pancytopenia (a decrease in the number of red and white blood cells and platelets in the blood): Symptoms similar to anemia, plus repeated infections (of varying severity) and localized or generalized bleeding.

Because of the risk of developing these issues, people with Seckel Syndrome should consider having regular blood tests. If symptoms appear, further tests should be performed.

TREATMENT:

The management of Seckel syndrome is multidisciplinary (surgery, occupational therapy, orthopedics, child psychiatry, physiotherapy, etc.) and preventive, and is aimed primarily at detecting complications and enabling those affected to have a better quality of life.

Currently, there is no specific treatment for Seckel syndrome. Tests on growth hormones show only a very limited effect. Limb lengthening, a controversial practice, is not recommended. In addition to the complications and pain it can create, this technique would create a body imbalance in people with Seckel syndrome.

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

- Clubfoot;
- Scoliosis or kyphoscoliosis;
- Intellectual disability;



- Strabismus;
- Blood disorders.

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 – fact sheet on Seckel syndrome
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Expert=808&lng=en

NORD - National Organisation for Rare Disorders
https://rarediseases.org/rare-diseases/seckel-syndrome/

GARD – Genetic and Rare Disease Information Center
https://rarediseases.info.nih.gov/diseases/8562/seckel-syndrome

OMIM - Online Mendelian Inheritance in Man
https://www.omim.org/entry/210600

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