

LÉRI-WEILL DYSCHONDROSTEOSIS

DESCRIPTION :

Léri-Weill dyschondrosteosis is a constitutional bone condition. In 70% of cases, it is due to a haplo-insufficiency (lack) of the SHOX gene. This gene, located on the sex chromosomes (X and Y), is responsible for growth regulation. In adulthood, the average height of affected individuals is 145 cm (4'9") in females and 155 cm (5'2") in males. The short stature is due to shortened limbs.

The prevalence of this diagnosis is estimated at 1 birth per 1000. It is transmitted in a pseudo-autosomal dominant mode, (i.e., each child born to an affected individual has a one in two chance of inheriting the condition). On the other hand, if both parents have dyschondrosteosis, the children have a one in two chance of having this type of dwarfism, a one in four chance of having another more involved form (mesomelic Langer's disease) and a one in four chance of not being affected [see the "genetics" sheet].

CHARACTERISTICS AND DIAGNOSIS :

Dyschondrosteosis causes a disproportionate type of dwarfism, affecting mainly the limbs. Expression of the diagnosis is variable, but clinical signs are generally more severe in women. Men have an athletic physique due to muscle hypertrophy without underlying muscle disease.

The main characteristics of the condition are as follows:

- Mesomelic shortening of the limbs (affecting the forearms and legs, below the knees);
- Madelung deformity (malformation of the wrists and forearm);
- Scoliosis;
- Shortening of the 4th metacarpus (hand bones);
- High palate.

Shortness of stature is present from birth, but because it is moderate, the diagnosis is often delayed and made during late childhood or early adolescence.

The diagnosis of Léri-Weill dyschondrosteosis is based on clinical and radiological findings and can be confirmed by molecular analysis. X-rays reveal, among other things, the shortness and irregularities of the forearm bones. The radii and ulna are short and arched; the lower end of the ulna has a characteristic "fork-back" shape, which results in subluxation of the wrists.



MAIN POSSIBLE COMPLICATIONS :

Joints are weak and range of motion is limited, especially at the knees, wrists and hips. Physiotherapy exercises can help improve mobility.

The Madelung deformity causes pain in the wrists and limits mobility. Clinical and radiographic follow-up should be done regularly to monitor the possible progression of the joint disease.

Splints can be used to reduce wrist discomfort.

Surgery (osteotomy of the ulna and radius with excision of the radioulnar ligament) may be necessary to alleviate pain and restore wrist function.

Scoliosis (three-dimensional deformity) may occur. Clinical monitoring should be performed regularly. It must be accompanied by x-rays as soon as any doubt arises. If scoliosis is detected, a corset or brace may slow further exaggeration of the spinal curvature. In some cases, surgery will be necessary.

TREATMENT:

The management of dyschondrosteosis is essentially limited to the treatment of functional restrictions and pain related to the diagnosis. This is why it necessitates several specialists such as orthopedists, surgeons or physiatrists.

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

- Madelung deformity;
- Scoliosis.

With regard to short stature, growth hormone treatment is effective and allows people with dyschondrosteosis to gain additional centimeters of bone length. Finally, another controversial treatment option, limb lengthening, is used only very rarely.

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 dyschondrosteosis

https://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=EN&Expert=240



Please contact us for more information

Association québécoise des personnes de petite taille
6300, avenue du Parc, bureau 430, Montréal (Québec) H2V 4H8

Phone: 514 521-9671 • Fax: 514 521-3369

Website: www.aqppt.org • E-mail: info@aqppt.org

Avec la participation financière de

Office des personnes
handicapées

Québec 