DIASTROPHIC DYSPLASIA

DESCRIPTION :

The term "diastrophic", which means "tortuous" in Greek, refers to the contorted appearance of the limbs of people affected by diastrophic dysplasia. This constitutional bone disease is caused by mutations in the SLC26A2 gene (or DTDST), which plays an important role in the development of cartilage. In adulthood, people reach an average height of 120 cm (3'11").
This diagnosis affects approximately 1 in 100,000 births. It is transmitted autosomal recessively [see "Genetics"].

CHARACTERISTICS AND DIAGNOSIS :

The condition causes disproportionately severe dwarfism. The limbs, much smaller than the body, are bowed. The trunk is shortened by scoliosis. The skull and face are not affected, but the ears are low.
Among the main morphological characteristics are the following:

- Clubfoot;
- Malformation of the fingers, especially the thumb (abduction);
- Kyphosis (humpback);
- Scoliosis (sideways curvature);
- Limitation or hyperlaxity of the joints;
- Cleft palate (opening of the palate) in about one-third of cases (sometimes associated with cleft lip);
- Hypodontia (missing teeth due to a developmental problem) in about one third of cases.

The expression of this type of dwarfism is variable, with severe and moderate forms, which may be diagnosed later. However, diastrophic dysplasia is often identified at birth by the discovery of micromelia (smallness) of the limbs, clubfoot, wrist deformity, and the particular implantation of the thumbs (proximal implantation).

Radiological diagnosis reveals short and massive tubular bones, enlarged metaphyses (growth plates located under the head of the bone), short and ovoid first metacarpals (hand bones), subluxation of the thumb (hitchhiker's thumb) and subluxation of the cervical vertebrae.

MAIN POSSIBLE COMPLICATIONS :

Development: There are no brain abnormalities, no risk of hydrocephalus (fluid accumulation in the brain) and no significant risk of cognitive abnormalities. However, limb and hand deformities cause delays in motor development and fine motor skills. Thus, the average age of

gait acquisition is about twenty-four months. Physiotherapy exercises can support a child's development.

Cysts: In the first few months of life, benign cysts of the outer ear appear in most infants. To remove them, a cast is preferred over drainage (surgery).

Cleft palate (opening of the palate): This malformation can lead to difficulties in swallowing and speaking. It can also cause middle ear dysfunction, so the ears and eardrums of infants with this issue should be monitored as early as nine or twelve months of age, and at regular intervals thereafter.
Surgery is available to correct cleft palate.

Clubfoot: Clubfoot is often challenging to regular surgery, and several operations may be required to correct it. For this reason, treatment should begin in the first few days of life. It includes softening and straightening of the foot, and the application of bandages and then plaster casts. Thereafter, the child will have to wear custom-made orthopedic shoes.

Knees and hips: Knee and hip mobility should be monitored in infancy and early childhood. In addition, an orthopedic and radiological assessment should be done in the first year of life and at least once a year thereafter.
Total hip and knee arthroplasty (surgery to restore a damaged joint) can be performed successfully in adults.

Scoliosis or kyphoscoliosis (a combination of kyphosis and scoliosis): About seven out of eight people with diastrophic dysplasia have this type of deformity. Scoliosis or kyphoscoliosis occurs very early, often in the first two years of life. It is important to address it as soon as the first manifestations appear.
A clinical evaluation should therefore take place every six months and should be accompanied by x-rays as soon as any doubt arises.
Treatment is difficult. Corset or brace interventions are generally insufficient and can only delay surgery. It is recommended that surgery be postponed as much as possible; ideally, it should be performed when the child is fully grown.

Spinal cord compression: This occurs in approximately 11% of people with diastrophic dysplasia. This problem results from the narrowing of the spinal canal, which compresses the spinal cord and can then lead to neurological problems.
Symptoms to watch for: Muscle weakness in the limbs, numbness, decreased fine motor skills, disturbances in walking and balance, functional impotence, and sphincter disorders (incontinence, dysuria, pollakiuria, and sometimes stress incontinence). These disorders, more common in people with scoliosis, usually appear in late adolescence.
As a preventive measure, a magnetic resonance imaging (MRI) test should be performed annually. If compression is detected, decompression surgery may be recommended.

Respiratory problems: Tracheomalacia and bronchomalacia are common in children. These problems affect the expiratory phase of breathing. Breathing is then wheezy or noisy, causing rapid shortness of breath. Infants may have episodes of cyanosis (blue discoloration of the skin).
Assessment of respiratory status should be done in early childhood. If wheezing increases and the child is experiencing respiratory distress, a pediatric pneumologist should be consulted.

TREATMENT:

The management of diastrophic dysplasia is multidisciplinary (surgery, orthopedics, occupational therapy, physiotherapy, etc.) and preventive, with the primary goal of detecting complications. Careful monitoring of spinal deformities (scoliosis or kyphoscoliosis) must be carried out. Currently, there is no specific treatment for diastrophic dysplasia. As an intrinsic bone and cartilage abnormality, treatment with growth hormone is not effective in treating this type of dwarfism. Finally, limb lengthening, a controversial practice, is used only very rarely.

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

- Developmental delays;
- Cysts (ears);
- Cleft palate;
- Clubfoot;
- Knee and hip degeneration;
- Scoliosis or kyphoscoliosis;
- Spinal cord compression;
- Breathing problems.

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 diastrophic dysplasia

https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=FR&Expert=628

Little people of America - Diastrophic Dysplasia Fact Sheet
https://www.lpaonline.org/assets/documents/NH%20diastrophic%20dysplasia1.pdf

Association Québécoise des Personnes de Petite Taille

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