MULTIPLE EPIPHYSEAL DYSPLASIA

DESCRIPTION :

Multiple epiphyseal dysplasia is a group of constitutional bone diagnoses that affect the epiphyses (the heads of bones). They are classified into six different types.

- Type 1 (or Fairbank type): Is an autosomal dominant trait/COMP gene mutation;
- Type 2: Autosomal dominant trait/COL9A2 mutations;
- Type 3: Autosomal dominant trait/COL9A3 mutations;
- Type 4: Autosomal recessive trait/SCLC26A2 mutations;
- Type 5: Autosomal dominant trait/ MATN3 mutations;
- Type 6: Autosomal dominant trait/COL9A1 mutations.

Adult height varies with diagnosis type but is generally between 145 and 170 cm (4'9" to 5'7"). Thus, most people are in the lower middle of the average growth curves, or just below.
Multiple Epiphyseal Dysplasia affects one in 10,000 or 20,000 births.

CHARACTERISTICS AND DIAGNOSIS :

Multiple epiphyseal dysplasia affects the ends of the long bones. The spine is rarely affected. Type 1 of the condition, often referred to as Fairbank type, is the most common and best known. There are syndromes associating epiphyseal dysplasia with other clinical manifestations such as myopia, deafness and facial dysmorphia.

The characteristics depend on the type of the diagnosis:

- Type 1: Mild short stature to average height, hips more affected, short hands and feet;
- Types 2, 3 and 6: Mild short stature to average height. More frequent abnormalities of the knees, milder and later joint involvement than in other forms, and in type 3 myopathy (muscle disease);
- Type 4: Slightly reduced adult height, characteristic double patella, clubfoot, irregularities of the hands, scoliosis, cleft palate (opening of the palate), joint stiffness;
- Type 5: Average height, sometimes myopathy (muscle disease).

Growth in early childhood often progresses normally, so the condition is diagnosed later, between 2 and 10 years of age, and sometimes in adulthood, when osteoarthritis begins to develop. During childhood, the first indicators of the presence of this diagnosis are a wobbly gait, limping, stiffness and joint pain (of the hips and/or knees in particular), increased fatigue during daily activities and difficulty running, as well as slower growth. Radiological examinations reveal bone dysplasia limited to the ends of long bones (epiphyses). In general, epiphyseal maturation is delayed and irregularities of the epiphyses are noted on x-rays.



PRIMARY POSSIBLE COMPLICATIONS :

Osteoarthritis: It is generalized and develops particularly in the knees, hips and shoulders. It is early and appears in adolescence or early adulthood.
Clinical and radiological monitoring must be carried out periodically from the time of diagnosis and tailored to the evolution of pain and functional limitations.

Hips: Hip degeneration and premature osteoarthritis are almost uniform and often occur in early adolescence. A significant portion of people with multiple epiphyseal dysplasia will undergo a total hip replacement around the age of 30 or 40.
Care must be taken, as hip involvement in late childhood may be reminiscent of Legg-Calvé-Perthes disease (necrosis of the femoral head), a more common condition.

Leg bowing and knee wear: Varus (outward) or valgus (inward) bowing may occur.
Leg alignment, chronic knee pain and limited walking should be monitored clinically.
Surgery should be reserved for patients with severe and symptomatic misalignment.
In addition, early osteoarthritis can cause premature wear and tear of the knees. Thus, a large proportion of people with multiple epiphyseal dysplasia will need to have their knees replaced around the age of 30 or 40.

Finally, **osteochondritis dissecans** (cartilage tears) may develop in the knees. In this case, it is necessary to stop all activities and immobilize the knees. If cartilage fragments are free in the joint, surgery is necessary. Rehabilitation (physiotherapy) must also be carried out.

TREATMENT:

The management of Multiple Epiphyseal Dysplasia is multidisciplinary (surgery, orthopedics, occupational therapy, physiotherapy, etc.) and preventive, and focuses on detecting complications. In particular, careful monitoring of osteoarthritis and joint degeneration must be carried out. Currently, there is no specific treatment for multiple epiphyseal dysplasia. As an intrinsic abnormality of bone growth, the use of growth hormones is not effective in treating this disease. 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"]:

- Osteoarthritis and degeneration of the hip and knees;
- Varus or valgus leg bowing.

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 multiple epiphyseal dysplasia
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=251

Little people of America - Multiple Epiphyseal Dysplasia Fact Sheet
https://www.lpaonline.org/assets/documents/NH%20Multiple%20Epiphyseal%20Dysplasia1.pdf

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