PROPOSAL FOR DIAGNOSTIC SUSPICION OF MUCOPOLYSACHARIDOSIS (MPS) IN PATIENTS WHO PRESENT WITH PATHOLOGICAL SHORT STATURE

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Introduction

PURPOSE : Short stature represents the main reason for consultation in Pediatrics Endocrinology . 20% of short stature is considered pathological and warrants further extensive studies.

A rare disease is one that have an incidence of less than 0.05% of the general population (1 in 20,000 people).Within which this the mucopolysaccharidosis.Severe cases may result in “Dwarfism” or significant growth impairment that may present after a period of normal development resulting in final heights below 3 to 6 standard deviations, hence the importance of an early diagnosis.

MATERIAL AND METHODS

:Literature review for clinical clues leading to consider bone dysplasias secondary to MPs in patients with pathological short stature

RESULTS 1

Consider MPs in patients with pathological short stature associated with:

1-Clinical presentation that compromise bone or cartilage normal growth and development (osteochondrodysplasia, dysostosis ,bone dysplasias as in skeletal deformity of the spine and/or extremities.

Spinal curvature severe abnormalities as kyphoscoliosis , genu valgum present after 18-24 months , pathological fractures, short extremities in the presence of anormal thorax.

Hand and feet distal epiphysis compromise, hands deformity , prominent and slightly wide knuckles. Flattening of Femur and tibia upper proximal epiphyses. Spasticity and non-congenital contractures, claw hand, nerve entrapment, restricted mobility and limited ambulation, hip dysplasia

RESULTS

.2- Progressive joint compromise without evidence of clinical or laboratory markers for inflammation.

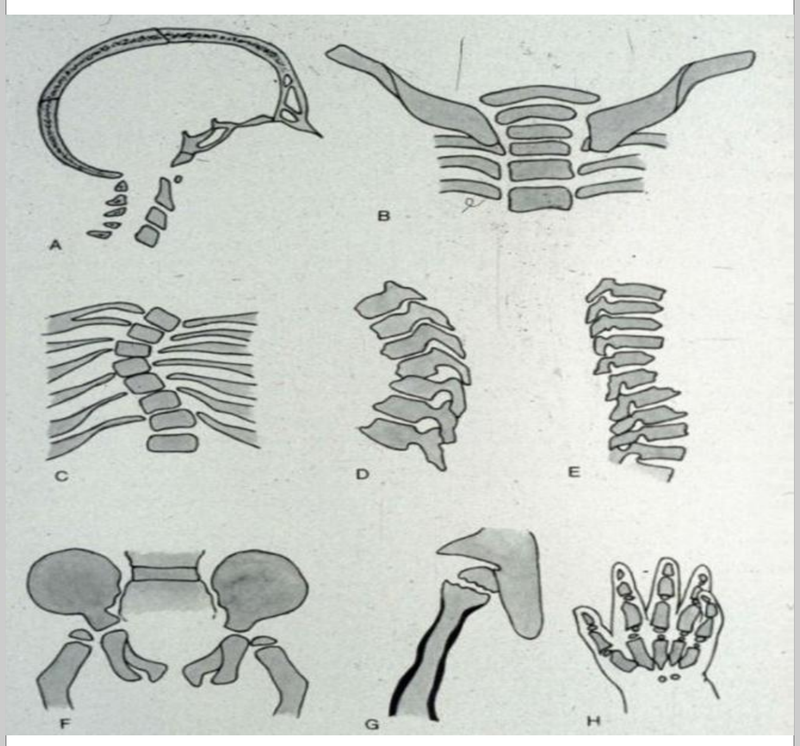
3- Progressive or degenerative character, marked slowing or loss of prior acquired skills, milestones.

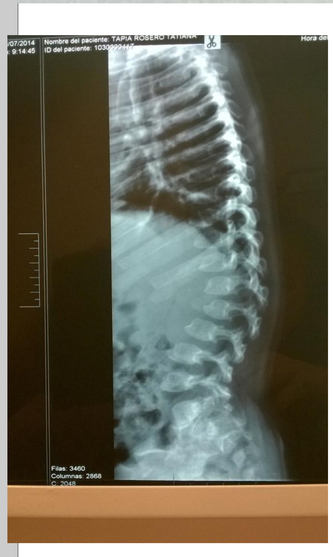
4- Evidence of visceromegaly or abnormal ocular signs

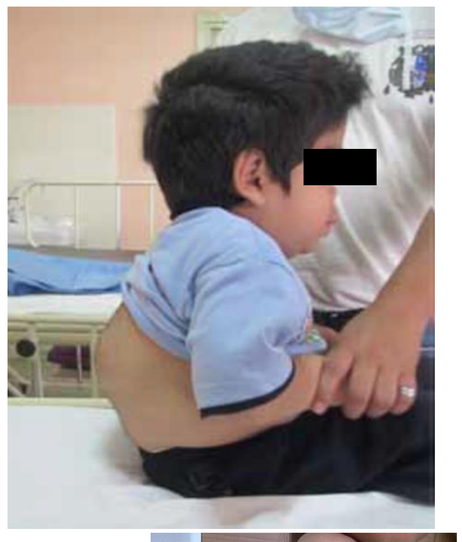
.5-History of hydrops fetalis or neonatal ascites

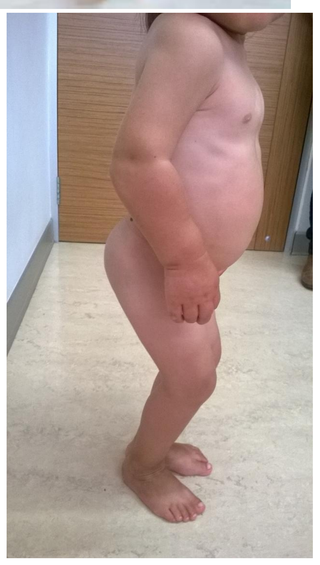
.6- Chromosomal abnormalities leading to primary calcium and phosphorus metabolic dysfunction (Rickets), and complex carbohydrates

PHOTOGRAPICS









CONCLUSIONS

Bone involvement leading to pathological short stature is due to underlying chondrocyte maturation disruption an location in the growth plate secondary to accumulation of MPs generating more compromise in axial growth than appendicular.

Short stature in MPs is secondary to a combination of metabolic, endocrine and skeletal structural abnormalities that restrict growth and final adult height.

Patients with attenuated forms might have normal or near normal lineal growth

.Other deposit diseases may share clinical features as those seen in MPs. Specific enzyme deficiency in leukocytes or cultured cutaneous fibroblasts will provide definite diagnosis.

Detailed clinical and radiological evaluation along with type of GAG eliminated in urine and leukocyte activity will narrow the differential diagnosis.

We proposed to look diagnostics MPS at patients with commitment to simultaneously of low sature compromide bone and facial.

Bibliografía

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