A RARE CASE OF GROWTH HORMONE DEFICIENCY MUCOLIPIDOSES TYPE II/III

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OBJECTIVES

Mucolipidoses II/III (ML) - rare autosomal recessive lysosomal storage disorders
- the range of inter- and intrafamilial variability, the variability of age at onset and organ manifestation is wide
- include growth retardation, facial dysmorphism, skeletal abnormalities, respiratory and heart diseases, hepatosplenomegaly , abdominal hernias.
The main objective of the present study was to conduct clinical analysis and treatment for a patient with MLII/III.

CASE PRESENTATION

- A.M., aged 18 y, boy of an apparently healthy couple
- first evaluation at 11y6m - short stature (-4 SD),
- coarse facial features,
- joint stiffness and pain initially in the shoulders, hips, and fingers
-thoracic deformity, kyphosis, clubfeet, deformed long bones
-cardiac involvement (insufficiency of the aortic valve)
- no signs of pubertal onset.

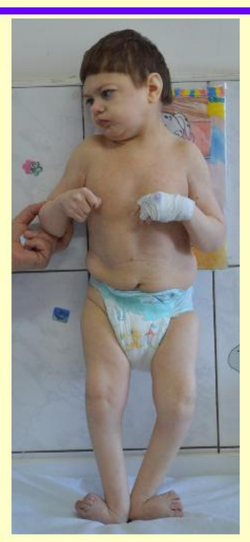

Fig.1

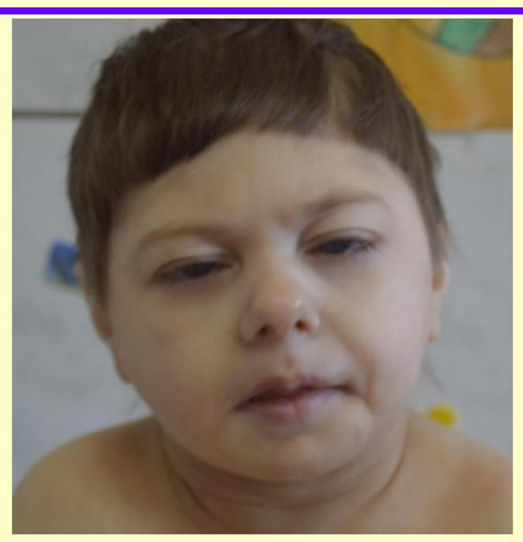

Fig.2

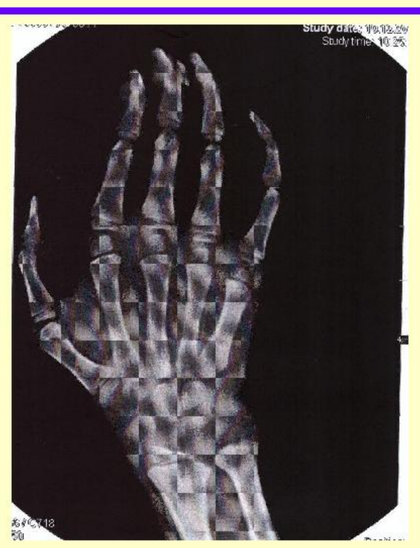

Fig.3

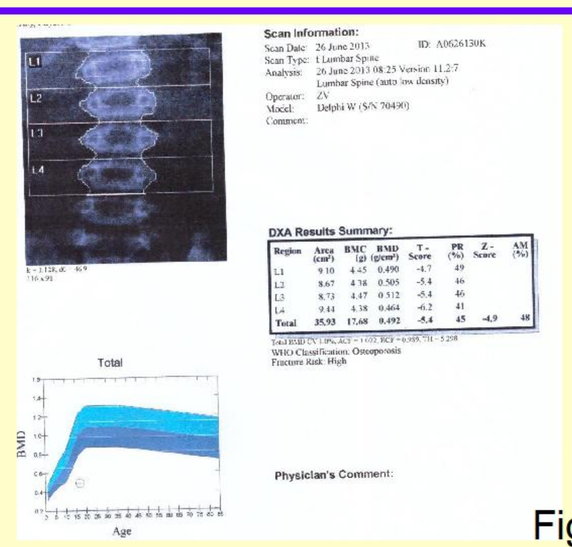

Fig.4

RESULTS

Wrist radiography - delayed bone age of 11 years 6 months (fig.3)
Somatotropic axis investigations - low IGF-1 =62.4 ng/mL, (N=220-972), GH=0.42 ng/mL, without stimulation at the arginine test: GH=2.75 ng/ml) → growth hormone deficiency.
Since there were not known contraindications, GH replacement therapy was started at age 11y 6m with an initial dose of 0.035mg/kg/day and biannual reassessments were performed.
After 4 years of treatment the medium growth rate was 0.42 cm/month and no side effects were reported.
Bone density. Generalized osteopenia - slowly progressive. (fig.4)
At the last evaluation the enzymes alpha-iduronidase, iduronate-2-sulfatase, arylsulfatase B, beta-galactosidase could be assessed and were higher in plasma → MLII or III.

CONCLUSIONS

Corroborating the clinical phenotype, biological data and evolution, this case can be included in MLIII.
We haven't found in the literature any case of MLIII treated with GH replacement therapy. In our case the treatment was effective and improved the patient's quality of life.

Later in the disease course management will be focused on relief of general bone pain associated with osteoporosis, which has responded in a few individuals to scheduled intermittent IV administration of the bisphosphonate - pamidronate.

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