

# ACCESS TO DIAGNOSIS AND TREATMENT OF PATIENTS WITH TYPE I MUCOPOLYSACCHARIDOSIS IN ROMANIA

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## Work hypothesis :

- Type I Mucopolysaccharidosis: - monogenic autosomal recessive disease  
 - induced by  $\alpha$ -L-iduronidase deficiency  
 ⇒ inability to catabolize GAG  
 ⇒ progressive cellular accumulation of GAG
- main characteristics
- craniofacial dysmorphism
  - visceral enlargement
  - cardiac complications
  - progressive mental retardation → dementia
  - skeletal anomalies
  - joint stiffness
  - severe somatic retardation
  - corneal clouding
- early diagnosis and treatment may improve the outcome

## Aim:

- assessment of therapeutical response to Aldurazyme in the first two romanian patients with type Hurler disease

## Material:

- two siblings with the diagnosis of MPZ type I, who have received enzyme replacement therapy for 18 months: - V.P., female, aged 9  
 - R. P., male, aged 4

## Work method:

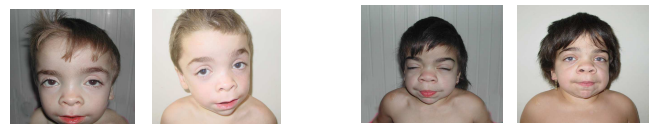
- the presumptive diagnosis of type I MPS was suggested by the characteristic clinical picture and was specifically confirmed by the dosage of leukocyte  $\alpha$ -L-iduronidase
- treatment with Aldurazyme in a dose of 1 mg/kg/dose was administered in i.v. perfusion, weekly, for 18 months
- the following parameters of the two children were monitored at baseline and after 6 months of treatment:
- clinical examination
  - somatometry
  - imaging examinations (radiological and ultrasonographic)
  - biochemical tests
  - functional respiratory tests
  - ophthalmologic
  - neurological and psychological examination

## Conclusions:

- Enzyme replacement therapy with Aldurazyme, which has recently become available for Type I MPS patients, has a favorable influence on the evolution and prognosis of these rare condition
- early diagnosis and treatment enables preventing irreversible damages
- Aldurazyme represents an extraordinary chance for type I MPS (Hurler disease) patients

## Results :

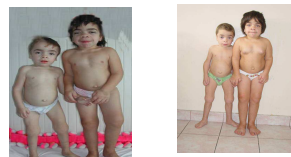
- 1) an improvement in craniofacial dysmorphism



R.P. before and after 18 months of ERT

V.P., before and after ERT

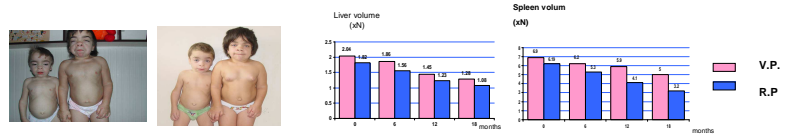
- 2) reduction in the flexion contracture of the joints of limbs with a better joint mobility; improved walking capacity



Before ERT

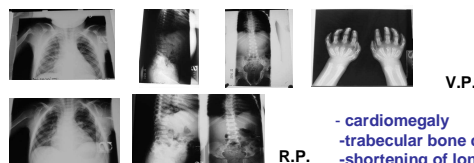
after 18 months of ERT

- 3) the liver and spleen volume decrease on ERT



The liver and spleen volum progression on ERT

- 4) bone impairment was maintained at the same level in both children on ERT



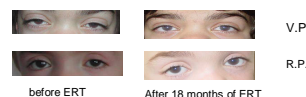
V.P.

R.P.

- cardiomegaly
- trabecular bone demineralisation
- shortening of long bones, pseudocystic aspect
- anterior platyspondilia, "beak shaped" vertebral bodies

- 5) moderate psycho-intellectual retardation in the girl, mild retardation in the boy did not progress.

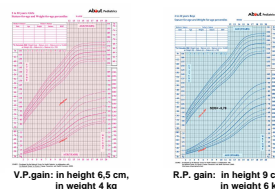
- 6) degenerative kerathopathy was maintained constant in V.P. and improved in R.P.;



before ERT

After 18 months of ERT

- 7) The statural growth was improved in both patients



- 8) The quality of life of the two siblings improved significantly.

