EARLY INFANTIL KRABBE DISEASE WITH UNUSUAL SURVIVAL

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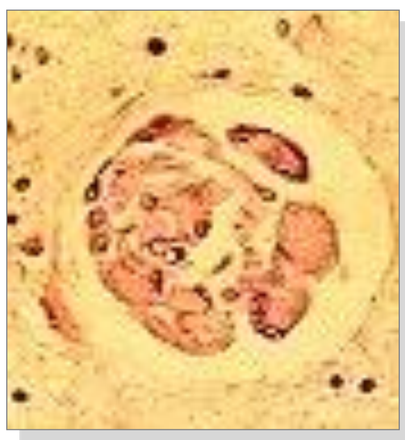
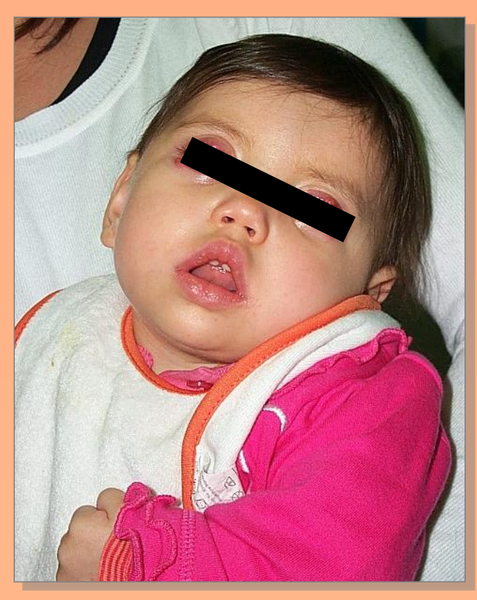
INTRODUCTION



- Globoid cell leukodystrophy (Krabbe disease) is caused by a deficiency of lysosomal galactocerebrosidase that results in progressive demyelination.
- The sole treatment is hematopoietic cell transplantation, which is only effective if performed before the onset of signs.
- In the absence of treatment, most children with early infantile Krabbe disease die within 2 years.

PERSONAL AND FAMILY HISTORY

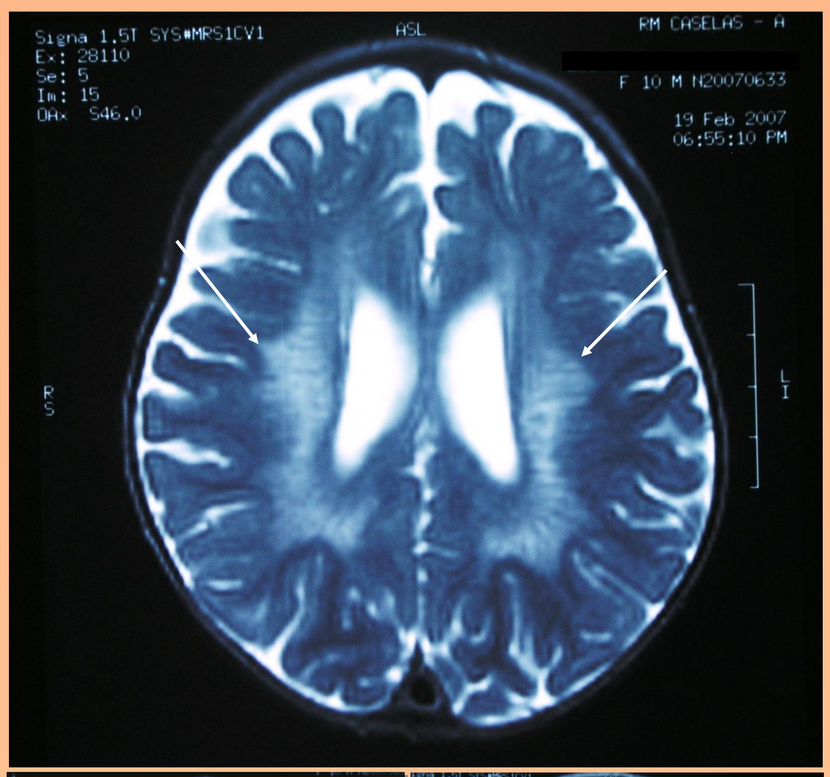
- LST., female, born on 21/4/2006
- 1st child of unrelated parents
- Normal pregnancy and term delivery. Apgar Score 9/9.
- BW 3.350g (P50), L 47,5cm (P10-25), HC 34,5cm (P50).
- Normal growth until 4,5 M of age
- Normal psychomotor development until 4,5 M of age



CLINICAL HISTORY

From 4,5 to 9 months of age:

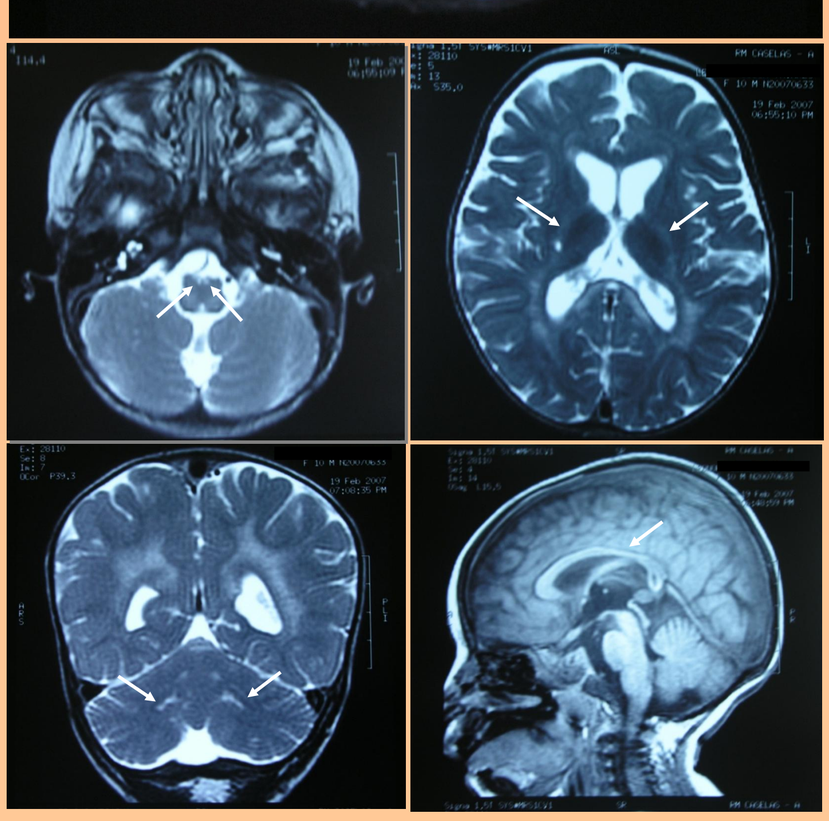
- Developmental delay → psychomotor regression
- Feeding difficulties → failure to thrive
- Irritability
- Stiffness with clenched fists
- Few spontaneous movements
- Spastic tetraparesia



DIAGNOSIS

- EEG : no paroxysmal activity
- Nerve conduction velocity: < 2 SD
- Magnetic Resonance Imaging: Extensive symmetrical cerebral white matter abnormalities with a tigroid pattern sparing the U-fibers
- MR Spectroscopy: ↓ NAA , ↑ choline, ↑ myoinositol
- Galactocerebrosidase activity*
  - Leucocytes: 1,06 nmol/h/mg protein (RV 2,18 – 22,4)
  - Fibroblasts: 0,43 nmol/h/mg protein (RV 3,54 – 24,1)
- Molecular study (compound heterozygosity)*
  - [GALC] del30kb; int10/end_del (intron 10)
  - [GALC] c.610 C>T; p.R204X (exon 7)

*(Enzymology Unit – Centro de Genética Médica Jacinto Magalhães)



EVOLUTION

- Since 9 months of age: no further progression of neurologic features
- Regression of irritability
- Good visual contact
- 5 years: W 6.000g, L 72cm , HC 45,5cm
- Frequent respiratory infections
- Death at the age of 5 years and 3 months

COMMENTS

- Our case shows that patients may have a stabilized form of disease and that a longer survival than described in the literature is possible without transplant in some patients.