ABOUT A CASE OF IDIOPATHIC DRAWFISM

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Objectives:

To report a case about Taybi linder syndrome wich is very rare cause of dwarfism
Taybi linder syndrome or microcephalic osteodysplasic primordial dwarfism type 1 (MOPD1) is characterized by an intrauterine and postnatal growth retardation , multiple malformations especially brain abnormalities.
the prognosis is poor with most of the reported patients dying within the first year of life

Methods:

We report a case of a boy, 2 years and 3 months with a neonatal history of intrauterin retardation (birth weight 1700g) consults for stunting. At birth, constataion of a microcephaly , many investigations were made but no diagnostisis established.
At the clinical examination we found a dwarfism(– 6,1 DS) (figure 4), microcephaly(occipital frontal circumference -7 DS)
Receding forhead, spars hair and eyerbrows, short neck (figure1), protruding eyes, small and low-set ears (figure 2) , postaxial polydactyly and bilateral cryptorchidism (figure 3).
Dry skin and genu varum; Delay of psychomotor acquisitions
MRI: partial agenesis of corpus callosum, lissencephaly, diffuse parenchyme atrophia
Radiographics: bone dysplasia: short necf of femur, enlarged metaphyses, absence of epiphyses(figure 5).

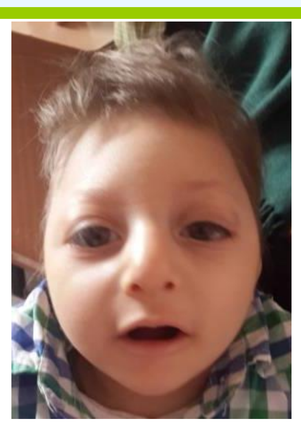


Figure 1

Figure 2

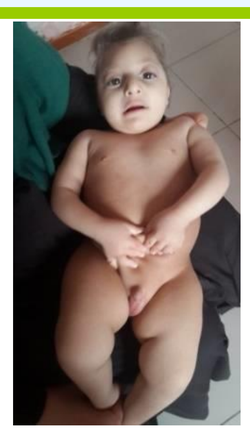


Figure 3

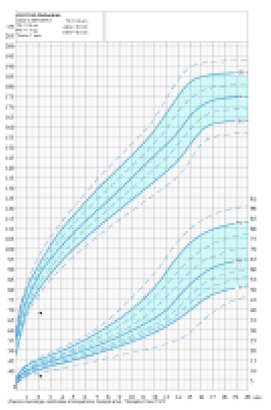
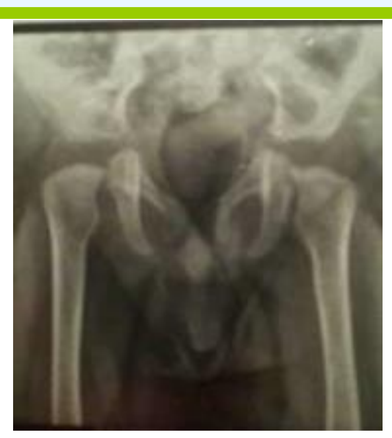


Figure 4



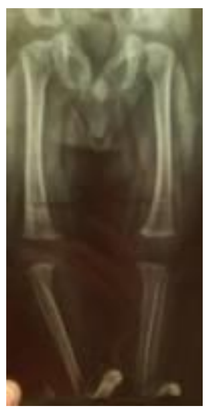


Figure 5

Results:

•On the basis of cliniacl and radiological phenotype ,the diagnosis made is microcephalic osteodysplasic primordial dwarfism type 1 for wich there is no treatment.
• A genetic study would be done at the genetic departments of Necker hospital but unfortunatly the patient died at the age of 2 years and 8 months.
• Microcephalic osteodysplastic primordial dwarfism (MOPD) type1 (OMIM 210710)was recognized 45 years ago by the description of two siblings with dwarfism, skeletal abnormalities and brain malformations by Taybi Linder (1967) . The syndrome was further delineated by Majewski and al(1982) as MOPD type I . Further mojor characteristics are extreme prenatal and postnatal growth retardation, severe microcephaly, unusual face and early death. The prevalence is unknown but less than 30 cases have been described in the literature.
• Mutations in the RNU4ATAC gene cause microcephalic osteodysplastic primordial dwarfism type I. It encodes U4atac, a small nuclear RNA that is a component of the minor spliceosome. Six distinct mutations in 30 patients diagnosed as microcephalic osteodysplastic primordial dwarfism type I have been described.
• The exact mechanisms by which decreased levels of spliceosomal complex RNAs might lead to the MOPD I phenotype remain unclear. Clues may be found by looking at other diseases caused by defects of the minor spliceosome. Although MOPD I is the first disease known to be associated with a defect in small nuclear RNA, it joins two other disorders defects in spliceosomal function – autosomal dominant retinitis pigmentosa and spinal muscular atrophy (SMA).
Treatment is supportive , prognosis is poor with most of the reported patients dying within the first year of life.

Conclusions:

Taybi linder syndrome or microcephalic osteodysplasic primordial dwarfism type 1 (MOPD1) the causative gene remains unknown but a mutation U4atac snRNAa component of the minor spliceosome in the developmental disorder.
Pronosis is fatal with death in the first or second year like our patient

References:

1- DU MOC 2014-2015
2- ORPHANET
3- Microcephalic Osteodysplastic Primordial Dwarfism type I with biallelic mutations in the RNU4ATAC gene Rebecca Nagy1,* , Heng Wang3, Beate Albrecht4, Dagmar Wieczorek4, Gabriele GillessenKaesbach4,5, Eric Haan6, Peter Meinecke7, Albert de la Chapelle1,2, and Judith A. Westman1 Clin Genet. 2012 August ; 82(2)
4- **Further delineation of the clinical spectrum in *RNU4ATAC* related microcephalic osteodysplastic primordial dwarfism type I** Ghada M.H. Abdel-Salam[1,*], Mohamed S. Abdel-Hamid[2], Nihal A. Hassan[3], Mahmoud Y. Issa[1], Laila Effat[2], Samira Ismail[1], Mona S. Aglan[1] and Maha S. Zaki **American Journal of Medical Genetics Part A** Volume 161, Issue 8, **pages 1875–1881**, **August 2013**

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