A HOMOZYGOUS DELETION OF EXON 7 IN LIFR GENE IN A NEONATAL CASE OF STUVE-WIEDEMANN SYNDROME

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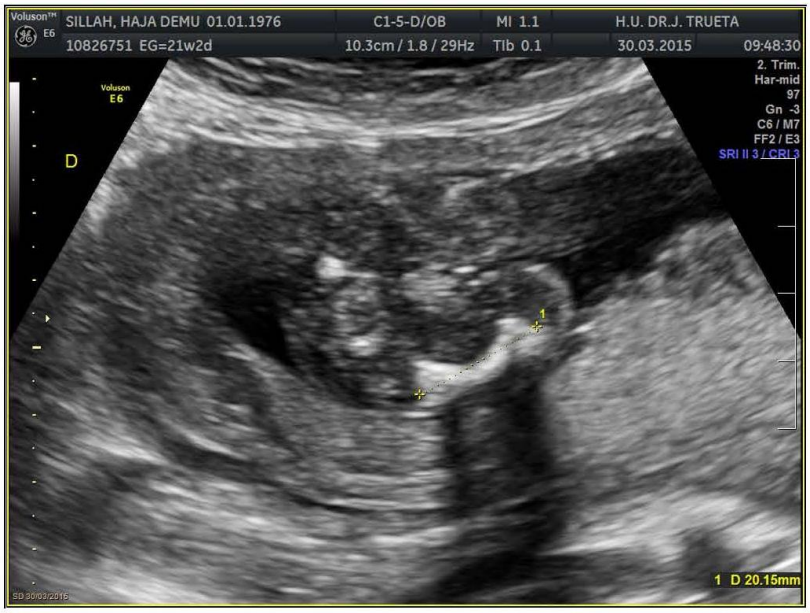
Abstract Body

INTRODUCTION: Stuve-Wiedemann syndrome (STWS; OMIM #610559) is a rare autosomal recessive bent-bone dysplasia characterized by bowed long bones, respiratory distress, feeding difficulties, and hyperthermic episodes. It was thought to be a lethal condition but there are reports describing patients who survive.
METHODS: We reported two sibs, with neonatal STWS characteristics, offspring of first-cousin parents heterozygotes for deletion of LIFR exon 7. In our Prenatal Unit, presumptive ultrasound diagnosis of skeletal dysplasia was given during the second trimester of pregnancy: it was revealed a fetus with evident bowing and shortening of the lower limbs, mainly femur and tibia, camptodactyly and signs of intrauterine growth restriction.
Parents, first cousin marriage, with positive family history of sib recurrence with a previous 8-month-old son dead with bone dysplasia and acute exacerbation of chronic shortness by bronchiolitis, refused prenatal diagnosis.
Male born at 42 weeks gestation after induced labor for oligohydramnios and with suspicion of skeletal dysplasia in utero.

RESULTS: At birth, clinical examination was concordant with the ultrasound findings. Parental consanguinity, sib recurrence and clinical findings suggested a STWS syndrome. Patient survives at 18 months with an improvement in their ability to swallow and regulate breathing as literature describes. Sequence analysis of LIFR gen was performed and there was no amplification of *exon* 7. An homozygous deletion was suspected and cDNA Sanger sequencing confirmed the homozygous exon 7 deletion [r.562_736del];[r.562_736del]. Variant does not describe to our knowledge at this time. Heterozygous carrier status of both parents was confirmed.

CONCLUSIONS: We underline the usefulness of ultrasound study of fetal skeleton in the prenatal diagnosis. The analysis supported the clinical homogeneity of SWS, despite genetic heterogeneity. Although STWS is a rare condition and the prognosis is poor, management strategies could increase long-term survival. Unfortunately, at the moment the family doesn't accept any genetic advice.

Abstract image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