From Ultrasound to Genotype:
PERIVENTRICULAR NODULAR HETEROTOPIA

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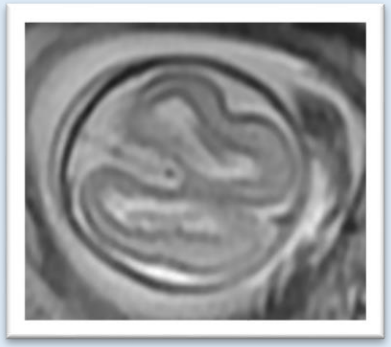
INTRODUCTION:

Periventricular nodular heterotopia (PVNH. OMIM # 300049) is a Dominant X-linked disease caused by a mutation of the Filamin A (FLNA) gen, in chromosome Xq28. PVNH is a neuronal migration disorder. Nodules of ectopic placed neurons are created. It is a heterogeneous and rare disorder that involves mainly neurological and cardiovascular manifestations. Usually females present the disease while males die as previable fetuses.

CASE REPORT:

25-year-old patient, G1, with no relevant medical history. Neurological and cardiological fetus alterations were detected in the regular 20 weeks ultrasound (U/S). Therefore, complementary tests were carried out: echocardiography, neurosonography and fetal magnetic resonance (MR) [Images]. They provided a better morphological diagnose:

-Bilateral periventricular nodular subependymal heterotopias
-Bilateral ventriculomegaly and Megacistern magna
-Artopulmonary disproportion and Atrioventricular valve dysplasia



Images 1 and 2: MR 23 weeks

Image 3: MR 26 weeks

The patient refused amniocentesis. Therefore, the genetic diagnosis could not be made until postpartum. Eutocic labor occurs at 40th weeks of gestation; being born a female of 3160 g (p33).
The newborn was asymptomatic at birth and she has not presented seizures so far.
She was heterozygous for FLNA gene variant mutationc.7898_7900delGGG (p.Gly2633del;NM_001110556.2).

Image 4: U/S 22 weeks

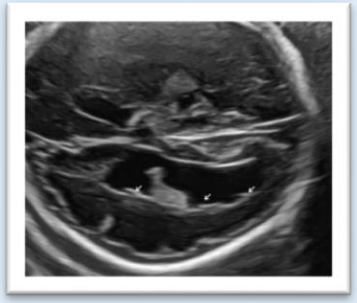


Image 5: U/S 29 weeks