XLAG SYNDROME CASE ACCOMPANYING A NEW ARX MUTATION AND HAS A INTERHEMISPHERIC CYST

S. Korkut[1], S. Doğanay[2], Ahmet Özdemir[1], Serkan Kurtgöz[3], Selim Kurtoğlu[1], Tamer Güneş[1]

Erciyes University School of Medicine, [1]Department of Neonatology, [2] Department of Pediatric Radiology, [3] Süleyman Demir University Department of Medical Genetic, Kayseri, Turkey

Background:

XLAG (X-linked lissencephaly with ambiguous genitalia) syndrome which is a clinical spectrum of ARX mutations is presented with severe growth deficiency, abnormal genitalia and resistant seizures in neonatal period. We present a XLAG case which was formed due to a new ARX mutation and has an appearance of a huge interhemispheric cyst different from classic neuroradiological imagings.

Case: The case which was diagnosed with prenatal hydrocephalus and had prematurity, low birth weight and respiration problem was taken to the newborn intensive care unit. In the physical examination, hypotonic, spontaneous movements were very few in macrocephalic appearance and the patient had highly arched palate and the face was dysmorphic (hypertelorism, micrognathia, long filtrum, thin upper lip, low ear). Fallus height was measured 1.1 cm in the genital inspection, ventral hypospadias was present and the gonads were inpalpable. Starting from the postnatal second day, cyclic movements in facial muscles and multifocal clonic seizures began. Serological tests were negative when TORCH infections were considered. Uterus and gonad were not observed in pelvic ultrasonography. Corpus callosum agenesis, gigantic interhemispheric cyst, lissencephalia and olfactory gyrus absence were detected in brain magnetic resonance imaging (Figure 3-5). Karyotype analysis 46XY was found. A new homozygote p.Thr357Asnfs*175 (c.1068_1069dupA) mutation was detected in DNa sequence analysis.

Conclusion: While karyotype is 46XY in ARX mutations, external genital abnormalities can change between hypoplastic penis or undescended testicle and complete female appearance. So genital examinations of lissencephalic cases should be made carefully, clinical spectrums of ARX mutations should be considered in the presence of indefinite genitalia.

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Gonads & DSD

Ahmet Ozdemir

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