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CASE 2017-6

Submitted by:

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Clinical History: A 35-year-old G3P2 at 20w5d of gestation was referred for Maternal-Fetal Medicine consultation. A genetic sonogram revealed multiple congenital anomalies including dolicocephaly, elongated and parallel lateral ventricles, choroid plexus cysts, nuchal thickening, frontal bossing, midfacial hypoplasia, hypertelorism, bell-shaped chest, multiple echogenic intracardiac foci, and bilateral clubfeet. X-ray examination prior to autopsy showed absent mid-phalanx of 5th finger of hands, clefted lumbar vertebral bodies and 11 ribs. A male fetus was delivered vaginally at 22w5d of gestation.

Autopsy findings: Body length: 32 cm, Body weight: 526 g; Brain weight: 154.3 g; Right and left feet length were 2.9 and 3.4 cm respectively. Autopsy examination confirmed imaging findings and revealed vascular malformations in various organs and brain abnormalities (brain- figure 1, coronal section of brain- figure 2)

Material submitted: Images of brain, including coronal section, and H&E sections of the cerebrum

Points for discussion: 1. Neurological manifestations of this syndrome
2. Pathogenesis and genetic pathway