Clinical Quiz

What is the Diagnosis?

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Case History

This case was initially referred to the genetics clinic by the obstetrician at 20 weeks of gestation in view of abnormal ultrasound scan. Increased nuchal translucency (9 mm) and cystic hygroma was noted in 12 weeks and 16 weeks scan. Scan at 20 weeks showed cystic hygroma, subcutaneous edema, short long bones, small stomach, clenched hands and some narrowing of pulmonary artery. There is a high risk in Down syndrome screening of 1:2, subsequent chorionic villus sampling showed normal karyotype and aCGH. After discussion with the obstetrician and geneticist, the couple decided to keep the pregnancy without further genetic testing for RASopathies. Ultrasound at 35 weeks showed polyhydramnios, rhizomelic shortening, clenched hands, flat forehead, low set ears and suspected hypospadias.

The baby was delivered using low forceps at 37+4 weeks with BW 3480 gram (50%), BL 49 cm (10%), HC 34 (10-25%). No resuscitation is required. The baby was dysmorphic with the following features: frontal bossing, tall forehead, sparse eyebrows, anteverted nares, large mouth with thin upper lips, small palpebral fissure with ptosis low set and dysplastic ears. Other clinical findings include short proximal limbs, camptodactyly, submucosal cleft palate, grade I subglottic stenosis, ectopic anus, micropenis, failed BAEP bilaterally, and suspected GERD. EDTA blood was sent out for whole exome sequencing and the report came back negative.

Clinical photos of patient at birth, 6 months old and 2 years old are shown in Figure 1.

Figure 1 Clinical photos of the patient. (Consent for publication has been obtained)

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N.B. The Editors invite contributions of illustrative clinical cases or materials to this section of the journal.