

Case report of a rare Bosma arhinia microphthalmia syndrome (BAMS)

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Case: A primigravida with so far normal pregnancy presented to us in the 30th gestational week with polyhydramnios and intrauterine growth restriction. A detailed ultrasound scan confirmed polyhydramnios with an amniotic fluid index of 30 cm, growth restriction under the 5th percentile and an abnormal facial profile with nasal hypoplasia / aplasia.

Aside from the mentioned abnormalities, the fetus showed normal cardiac and neurocranial ultrasound findings. These findings remained throughout the rest of the pregnancy with increasing polyhydramnios and subsequently cervical insufficiency. Amniocentesis was performed to reduce the intrauterine pressure on one hand and to perform genetic diagnosis on the other. The karyotype was normal 46 XX, but further diagnosis with Microarrays was not performed as per wishes of the parents. A follow-up in the 38+4 week of pregnancy showed pathological cardiotocogram, so that a caesarean section needed to be performed.



Figure 1: Ultrasound finding of the facial profile

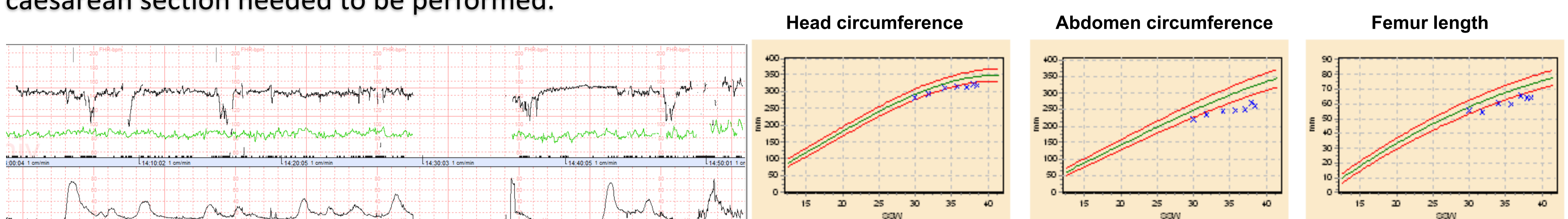


Figure 2: antenatal cardiotocogram and the fetal growth below the 5th centile

A hypotrophic baby girl of 1810g without a nose or nasal orifices was born. After securing an oropharyngeal airway, the newborn adapted well and showed normal heart rate and Oxygenation. Clinical assessment confirmed bilateral microphthalmia and colboma without further abnormalities. MRI showed that arhinia, microphthalmia, and colboma were the only anomalies. The baby was able to breathe normally through the mouth and even oral feeding could be started without complications.



Figure 3: the postnatal clinical presentation of BAMS

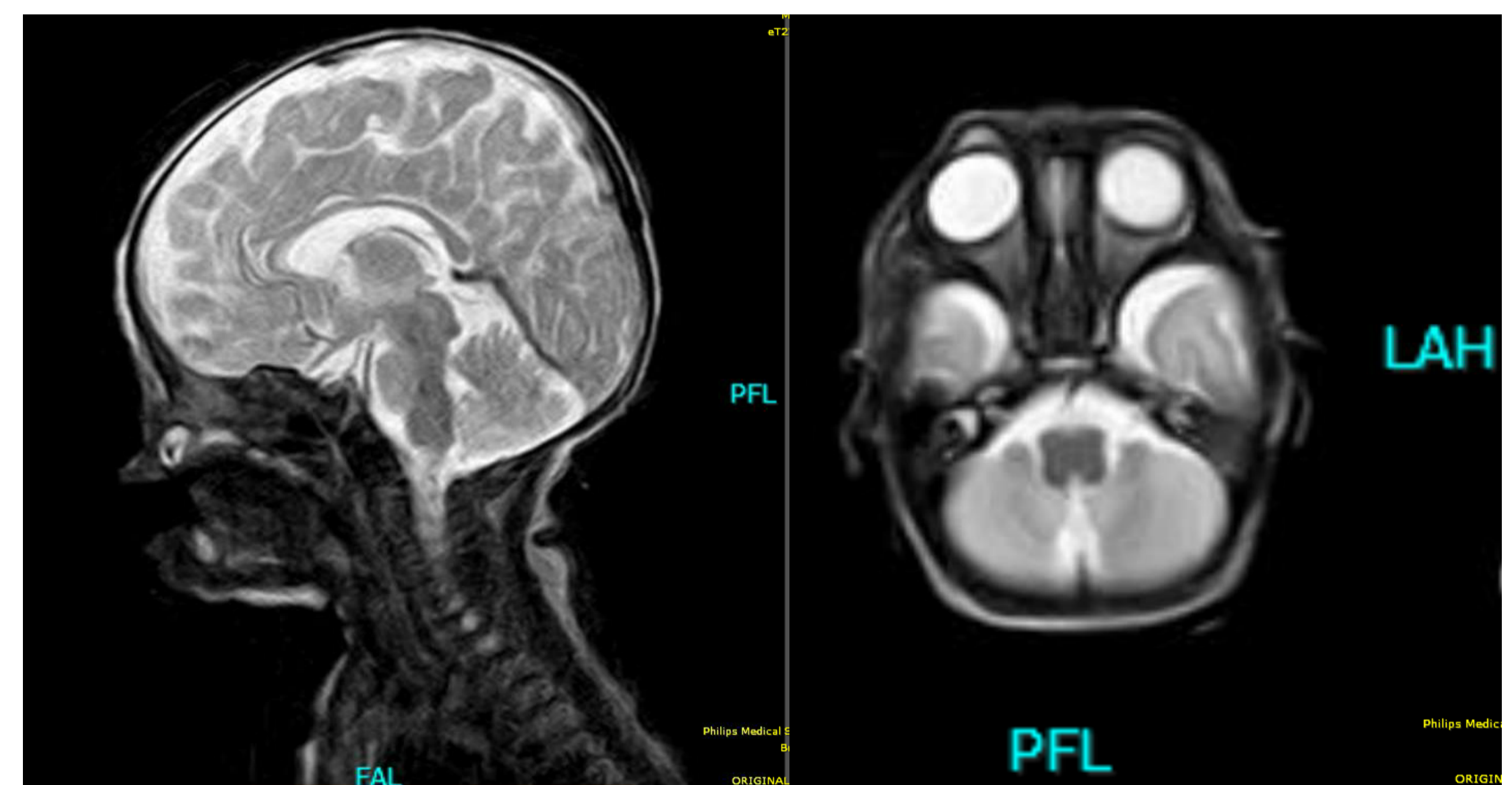


Figure 4: cranial MRI finding of BAMS

Discussion: Congenital arhinia is an extremely rare condition that describes the complete absence of the nose. This condition has been reported in around 50 cases in published literature, which shows how rare the incidence is. It could be isolated or associated with other abnormalities such as mid-face hypoplasia, nasolacrimal duct anomalies, high-arched palate, and ocular defects including hypertelorism, microphthalmia, and colbomas. Furthermore reproductive axis defects in males as well as absent olfactory bulb have been reported. Bosma arhinia microphthalmia syndrome is characterized by absence of the nose as well as unilateral or bilateral microphthalmia. Gordon et al 2017 published a series of 14 cases of patients with BAMS and was able to describe the genetic defect behind the syndrome. A de novo heterozygous missense mutation was identified in the epigenetic regulator of the SMCHD1 was found in all cases, thus established that the SMCHD1 gene is essential in the nasal development. Postnatal sequencing and genetic testing confirmed that our case had a mutation in the SMCHD1. This is valuable information, which confirms the findings of Gordon et al. and adds another patient to the published 14 cases.

Conclusion: This rare case confirms the mutation of the SMCHD1 gene as a cause for BAMS. Prenatal genetic testing for this mutation in cases of fetal arhinia can provide definitive diagnoses and improves prenatal counseling and perinatal management.