COULD ABNORMAL FETAL MOVEMENTS BE A CLUE TO THE PRENATAL DIAGNOSIS OF PONTOCERELLAR HYPOPLASIA AT MIDGESTATION?

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INTRODUCTION:
The pontocerebellar hypoplasias (PCH) are a group of early-onset, autosomal recessive disorders resulting in abnormal growth and function of the brainstem and cerebellum. Pontocerebellar hypoplasia type 1 (PCH1) causes problems with movement characteristic of spinal muscular atrophy, hypotonia, contractures, microcephaly and breathing problems that are evident at birth. Most children with PCH1 live only into infancy. Although this is a congenital disorder of pontocerebellar dysgenesis with fetal onset of neurodegeneration and symptoms at birth, no case of prenatal diagnosis based on prenatal ultrasound (US) or magnetic resonance (MR) imaging has been described yet.

CASE REPORT:
We report prenatal diagnosis of PCH1 in pregnancy referred to us at 22 weeks due to the abnormal family history. Older sibling died at the age of 8 months on dyskinesia with dystonia due to the complex congenital CNS defect (atrophy of caudal parts of cerebellar hemispheres and peduncles, brainstem and pons, megacisterna magna and hypoplasia of corpus callosum). No exact genetic origin of this disease was found thus molecular testing wasn’t possible. AR or X-linked inheritance was assumed with estimated risk of around 25%. Our ultrasound examination at 22 + 3 revealed polyhydramnios, abnormal (spastic) fetal movements and abnormal holding of fingers resembling clenched hands. CNS anatomy was normal both by US and MR imaging. The patient was referred by geneticists for second opinion examination to another fetal medicine specialists, who described isolated polyhydramnios with normal fetal growth, movements and anatomy. The mother decided to visit our centre at 27 + 3 again; result of this US examination was similar to that at 22 weeks. Another follow-up scan was recommended in about one moth.

We examined at 32 + 2 days revealed CNS abnormality, namely cerebellar hypoplasia. Polyhydramnios, abnormal fetal movements with abnormal holding of fingers were still present too. Fetal MRI at 33 + 2 days confirmed cerebellar hypoplasia affecting predominantly lower cerebellar hemispheres.

Figure 1: Prenatal and postnatal examination by US and MRI. (1a-c) prenatal ultrasound examination at 22 + 3 presenting abnormal holding of fetal hands and fingers; (1d) prenatal ultrasound examination at 32 + 2 showing cerebellar hypoplasia; (1e) fetal magnetic resonance imaging showing cerebellar hypoplasia at 33 + 2; (1f) postnatal magnetic resonance imaging (T1-weighted images) presenting cerebellar hypoplasia affecting mainly inferior parts of cerebellar hemispheres.

CONCLUSION:
Our case demonstrates the importance of observation of fetal movements during anomaly scan at midgestation, in particular in pregnancies being in high-risk of CNS defects. Although assessment of fetal movements is mainly subjective thus depends on operator’s expertise and experience, abnormal fetal movements should not be underestimated and detailed fetal brain examinations with prenatal genetic testing (micro-array, other tests) should be considered.