Joubert Syndrome diagnosed at 16+6 weeks gestation and Molar tooth sign by 3D modality

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Objective

Joubert Syndrome is an uncommon autosomal recessive neurodevelopmental disorder involving cerebellar vermis and brain stem. We report a case of a fetus presented on antenatal scan at 16 weeks + 6 days with characteristic Molar tooth sign.

Methods

28-year-old, G8 P5+2 had consanguineous marriage. GDM on diet. Previous child with Vermian Agenesis and Polyhydramnios. Has 4 other children who are healthy. Had consanguinity in the other family members and family history of recurrent fetal loss with hydrocephalus. She was referred to our fetal medcine unit for early anomaly scan at 16 weeks + 6 weeks due to previous history. On ultrasound characteristic molar tooth sign, absent cerebellum and vermis, frontal bossing with hypertelorism, dilated anterior horn of the lateral ventricle, enlarged cisterna magna with posterior encephalocele, kyphoscoliosis and bilateral renal pelvic dilatation. With these findings and history of previous affected child most likely diagnosis was made to be Joubert syndrome. Based on these findings parental counseling was made and opted for termination at 17 weeks. Sample from an aborted fetus was consistent with the diagnosis of Joubert syndrome type 21. Genome sequencing confirmed CSPP1 (NM_024790. 6; exon 17,) homozygous likely pathogenic variant c. 2131_2132del p. (Ser711Leufs*11).

Results

Prenatal sonographic findings are relatively nonspecific and include increased nuchal translucency (NT), enlarged cisterna magna, cerebellar vermian agenesis, occipital encephalocele, ventriculomegaly, hypoplastic phallus, renal cysts, cleft lip or palate, polydactyly and tongue abnormalities. Discussion: The disorder is characterized by absence or underdevelopment of the cerebellar vermis and a malformed brain stem (molar tooth sign), both of which can be visualized on a 3D USS/MRI scan Together with this sign, the diagnosis is based on the physical symptoms and genetic testing for mutations. Most of the signs and symptoms of appear very early in infancy with most children showing delays in gross motor milestones. Signs and symptoms vary the most common features include ataxia, hyperpnea, sleep apnea, abnormal eye and tongue movements, and hypotoniaThis disorder can be caused by mutations in more than 30 genes within genetic makeup.

Conclusion

Prenatal sonographic findings in fetuses with Joubert Syndrome are relatively nonspecific and include increased NT, enlarged cisterna magna, cerebellar vermian agenesis, occipital encephalocele, ventriculomegaly, hypoplastic phallus, renal cysts, and polydactyly. In the present case we were able to identify the molar tooth sign following in 4d scan. As the syndrome is associated with a bad outcome the pregnancy was terminated at 17 weeks. Early diagnosis is of great value in Islamic country as termination of a pregnancy is not feasible beyond 18 weeks GA. Previous history with USS findings helped in making early diagnosis. Carful history is important.