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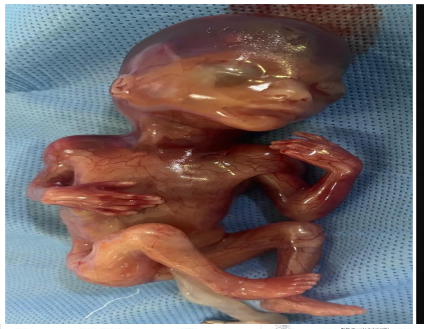
Title: A Rare Case of anomalous baby with Dandy Walkar and Joubert syndrome features

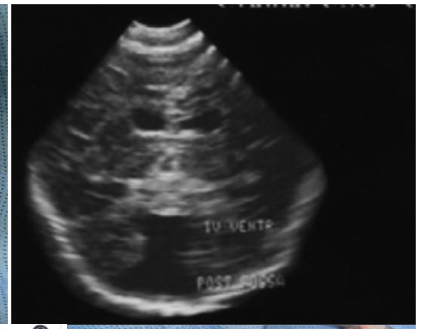
INTRODUCTION:

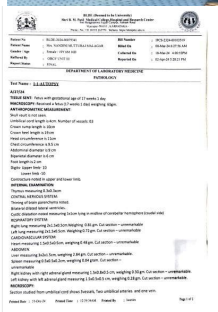
- Dandy walkar malformation(ZIC1 and ZIC4 genes) is characterised by hydrocephalus,partial or complete absence of cerebellar vermis and posterior fossa cyst found in 1:25000 live births,while Joubert syndrome(autosomal recessive) is associated with distinctive brainstem and cerebellar malformations affecting 1:80,000 live births with molar tooth sign on MRI/USG.If the findings of these diseases were found mixed in a fetus,identification of the cause of the anomaly can be tricky.(1)

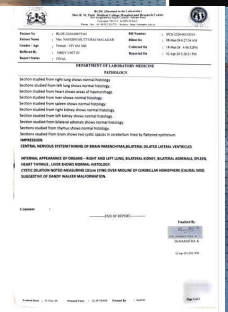
CASE DISCUSSION:

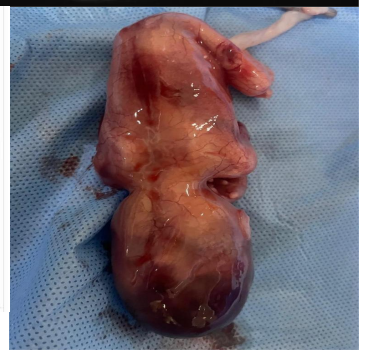
- A 18 year old female named X who is a primigravida with 2° consanguineous marriage with 17 weeks 1 day period of gestation has come for regular antenatal care.
- An early anomaly scan was done which showed enlarged bilateral ventricles,evidenced by floating or dangling choroid occupying less than 2/3rd of the ventricular cavities,thalamopeduncular elongation with an obtuse thalamopeduncular angle on axial images and tectal breaking of the midbrain on saggital images suggesting a molar tooth sign.
- There was also dilated 4th ventricle with an unusual posterior turn communicating with a cystic structure measuring 6.3x3.6mm lying in midline caudal to the occipital bone,cisterna magna was found to be obliterated with flattened cerebellum.
- Because of these anomalies in the baby, the couple was adviced for termination following which the abortus was sent for autopsy and karyotypimg was done for both the parents to store the blood sample.The skin and blood samples of the fetus were sent for complete genome sequencing.











autopsy report

- The autopsy suggested the diagnosis of Dandy walkar malformation but not of joubert syndrome.
- The karyotyping analysis was normal for both the parents and NPHP1 X linked recessive gene mutation of the study was adviced(2),the genetic sequencing couldn't be performed due to contamination in the tissue sample.So genetic sequencing is necessary to identify the cause and pinpoint the mutational cause of the disease.

CONCLUSION:

- Early diagnosis of such genetic defect and further evaluation of the parents for such genetic traits help the parents in decision making regarding the continuation or termination of the pregnancy and in educating them regarding the chances of bearing similar babies in future pregnancies.

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2. Pugash, D., Oh, T., Godwin, K., Robinson, A.J., Byrne, A., Van Allen, M.I. and Osiovich, H. (2011), Sonographic 'molar tooth' sign in the diagnosis of Joubert syndrome. Ultrasound Obstet Gynecol, 38: 598-602.