Case Report

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Fraser Syndrome: Case Report

Nosheela Javed[1], Zeenat Usman[2], Sara Usman[3]

[1]Professor & HOD, Dept of Obs and Gynae, [2]Medical Officer, [3]Post Graduate Resident PIMS/SZABMU, Islamabad

Address of Correspondent
Dr. Zeenat Usman
Medical Officer, PIMS/SZABMU, Islamabad
usmanbut2@gmail.com

ABSTRACT

Pakistan is a low resource country where the literacy rate is poor. Women scarcely receive antenatal care. We present a case of Fraser syndrome. The patient in question was referred by a lady health worker to the tertiary care hospital for workup at 7 months of gestation. The patient reported late she was clinically assed a multidisciplinary approach was taken. Radiologically the fetus had deranged Doppler umbilical artery and severe oligohydoamionos. Suspicion of Limb anomalies was also given. A consultant led obstetrical team decided on not to proceed with caesarean section but to opt for vaginal delivery. The baby was delivered breech and was anomalous and expired soon after birth. Fortunately, due to team work the patient was saved from unnecessary surgical intervention.

Keywords: Antenatal care, Fraser syndrome, Multidisciplinary approach.

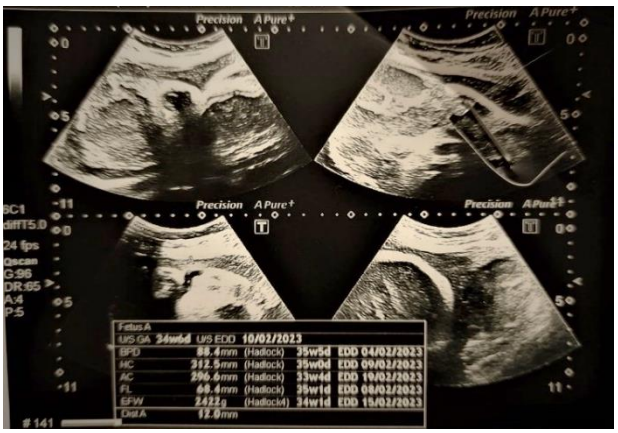
Cite this article as: Javed N, Usan Z, Usman S. Fraser Syndrome: Case Report. Ann Pak Inst Med Sci. 2024; 20(SUPPL-1):650-651. doi. 10.48036/apims.v20iSUPPL-1.1132

Introduction

Fraser syndrome is a rare genetic disorder that is an autosomal recessive malformation syndrome.[1] Its prevalence is 0,43:100,000 .This diagnosis is usually missed on prenatal ultrasounds due to rarity.[2] Various other names for it include Fraser-Francois syndrome, MeyerSchwickerath's syndrome, Ulrich-Feichtiger syndrome or cryptophthalmos-syndactyly syndrome.[3] This syndrome has a formulated diagnostic criteria that includes four major and eight minor features. Proper antenatal care should be given and such cases picked up before age of viability.[4] multidisciplinary approach is very much necessary for the benefit of the patient.[5]

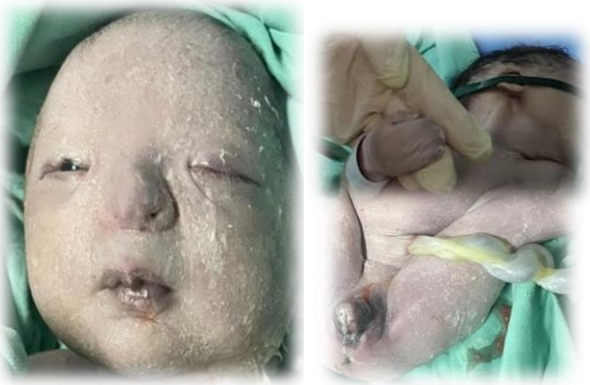
Case Report

A 29 year old lady gravida 5 para 2 and 2 miscarriages, unsure of dates as she conceived during lactational amenorrhea. She was married for 9 years to her cousin. Her first 2 pregnancies were spontaneous first trimester miscarriages' followed by two female off springs age 3 and 1 year respectively. She got to know she was pregnant when she got her ultrasound done at 26 weeks and that she had decreased liquor. She was referred by local lady health worker to PIMS in view of oligohydroamnios secondary to premature rupture of membranes. The patient did not give history of per vaginal leaking on examination the symphysio fundal height was smaller than the estimated gestational age, liquor was also clinically reduced and aminicator was negative. Radiologically severe oligohydroamnios and moderately deranged umbilical artery Doppler indices were given she was given antibiotics and asked to follow up for complete anomaly scan. Patient lost to follow up and reported at 9 months due to lower abdominal pain. On examination symphysio fundal height was 32 cm at term and fetus was breech. A gross anomaly scan was conducted that demonstrated severe oligohydroamnios AFI 1cm and intrauterine growth restriction. There was a suspicion of abnormalities but due to the immensely decreased liquor it was highly suboptimal study.



The decision to conduct a cesarean section was deferred. The patient went into spontaneous labour and an assisted vaginal breech delivery was conducted. Usually fetus with intrauterine growth restrictions are not given trials of

labour as they are unable to sustain the harsh conditions of labour but the consultant led team decided in favor of a normal vaginal. The baby turned out anomalous and expired soon after delivery. Diagnostic features of Fraser syndrome were identified cryptophthalmos-syndactyly and abnormal genitalia



Discussion

Fraser syndrome is very difficult to diagnose antenatally these cases usually present with severe oligohydroamnios secondary to renal agenesis.[2] Diagnosis is made on the basis of clinical features the most eminent of them being cryptohtalmos, that is a layer of skin replaces the eyelids and extends from the forehead to cheek with absent palpebral fissures. Ocular structures are poorly developed.[6] Other features include syndactyly, abnormal urogenitalia, deformities of nose ears, cleft lip, renal agenesis and mental retardation.[7, 8]

This syndrome is caused due to the genetic defect in FRAS1 and FREM2.This leads to failure of physiological apoptosis causing fingers to not form and palpebral fissure deformities or absence. GRIP1 and other genes encoding extracellular matrix proteins have also been identified as a probable cause of Fraser syndrome.[1] Whole exome sequencing is recommended in counsaigeous parents. In our country where genetic studies are extremely expensive.[9] An alternative is ultrasound which is an affordable modality. Prenatally it can be diagnosed in the hands of an expert operator.[10] The clinician should be well aware if such sonographyical picture is obtained and a multidisciplinary approach should be taken up.[11]

Conclusion

Genetic disorders are common in interfamily marriages hence a high index of suspicion should be kept. A good multidisciplinary approach with an affordable alternative can lead to less morbidity in patients [5,10]

Acknowledgements: We thank the patient for her cooperation, the doctors and staff of radiology, obstetrics and pediatrics for there contribution

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