Successful Term Pregnancy In A Woman With Noonan Syndrome: A Case Report

S Maiti

Citation

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Abstract

First described by Noonan and Ehmke (1963), Noonan Syndrome is an Autosomal Dominant condition with variable expression. It is associated with short stature, short neck with webbing (phenotypically like Turner Syndrome), cardiac anomalies particularly dysplastic pulmonary valve with Pulmonary stenosis(50%), atrial septal defect(10%), ventricular septal defect, patent ductus arteriosus, hypertrophic cardiomyopathy, a characteristic chest deformity with pectus excavatum inferiorly and pectus carinatum superiorly(70%), widespread nipple and hypertelorism. The chromosomes are normal.

We report a case of a 29 yr old primigravida with Noonan Syndrome sought preconceptional counselling and conceived within 6 months. Throughout the antenatal period , she was followed up closely by a team comprising of Obstetrician, Cardiologist, Ultrasonologist and her General Practitioner.

BACKGROUND

First described by Noonan and Ehmke (1963), Noonan Syndrome is an Autosomal Dominant condition with variable expression. It is associated with short stature, short neck with webbing (phenotypically like Turner Syndrome), cardiac anomalies particularly dysplastic pulmonary valve with Pulmonary stenosis(50%), atrial septal defect(10%), ventricular septal defect, patent ductus arteriosus, hypertrophic cardiomyopathy, a characteristic chest deformity with pectus excavatum inferiorly and pectus carinatum superiorly(70%), widespread nipple and hypertelorism. The chromosomes are normal.

CASE REPORT

A 29 yr old primigravida with Noonan Syndrome sought preconceptional counselling and conceived within 6 months. Throughout the antenatal period , she was followed up closely by a team comprising of Obstetrician, Cardiologist, Ultrasonologist and her General Practitioner.

On Echocardiography, mild pulmonary stenosis (pulmonary gradient of 17 mmHG) was detected. Cardiologist advised for antibiotic prophylaxis only for instrumental delivery, if needed for any obstetric reason..On referral to a tertiary genetic referral centre, a candidate gene for Noonan syndrome was found. In view of 50% risk of inheritance prenatal diagnostic option was discussed with its pros and cons. She declined the invasive prenatal diagnosis and wanted to go ahead with pregnancy anyway.

Fetal anatomy scan, detailed cardiac scan and serial growth scans were normal. She developed thrombocytopenia (86 109 /L) in 3rd trimester which recovered in the immediate postnatal period. She presented at 38 weeks with spontaneous labour. A live female baby of 3190 Gms (APGAR scores 8 1, 10 5) delivered by emergency lower segment caesarean section under epidural anaethesia for failed progress in first stage. She had an uncomplicated recovery and discharged home with baby on 4th post-operative day.

DISCUSSION

Noonan syndrome is characterised by late onset and progressive pathologies, particularly associated cardiac anomalies which continues to develop even in postnatal life. This complicates or precludes prenatal ultrasound diagnosis. [1]

As this is an Autosomal dominant condition, the inheritance rate is 50%. Earlier diagnosis will improve clinical management and genetic counselling. The mean age of diagnosis in the study conducted at St George' s Hospital, London with 151 patients with Noonan Syndrome was 9 yrs. [2] A well co-ordinated multidisciplinary team approach is important for a successful outcome in a pregnant woman

with this condition.

References

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Author Information

S. Maiti, MD, MRCOG, DFFP
Research Registrar, Wirral Hospital NHS Trust, Arrowe Park Hospital