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Case Report of Tuberous Sclerosis

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ABSTRACT

BACKGROUND:-Tuberous sclerosisis a rare**,** multisystemdisease (skin,brain,heart,kidney,eye,lung,bone) characterized by an autosomal dominant mode of inheritance. It is a type of neurocutaneous, genetic disorder. Prevalence of disease in newborn is 1 in 6000 to 1 in 10000 .Two main foci involved are TSC1 gene and TSC2 gene. Hallmark of the disease is the CNS involvement. 50% of Newborns had shown Cardiac Rhabdomyoma, detected antenatally by ANC USG at 20-30 weeks of gestation.

CASE REPORT :-

Patient was referred from Navsari Civil Hospital on first day of life for respiratory distress along with ANC USG suggestive of multiple rhabdomyoma in heart and tuberous sclerosis. Baby was kept on O2 hood for distress. Chest-x-ray done was suggestive of cardiomegaly ,2D-Echo done was suggestive of rhabdomyoma of heart.

DISCUSSION :Definite tuberous sclerosis diagnosis made when atleast 2 major or one major plus 2 minor feature are present. In addition, carrying a pathogenic mutation in TSC1 gene Or TSC2 gene is sufficient.

CONCLUSION: if antenatal USG misses out the diagnosis of tuberous sclerosis,one can confirm the same postnataly by 2D-Echo.

One should also be vigilant regarding cardiac,renal, neurological related complications.

KEYWORDS

Tuberous Sclerosis, Rhabdomyoma of Heart, Autosomal dominant, Lymphangiomyomatosis, sub-ependymal nodule.

I. INTRODUCTION:

Tuberous Sclerosis is a rare neurocutaneous, genetic disorder characterized by an autosomal dominant mode of inheritance. Also characterized by presence of congenital benign tumors in multiple organs (skin,brain,heart,kidney,eye,lung,bone).

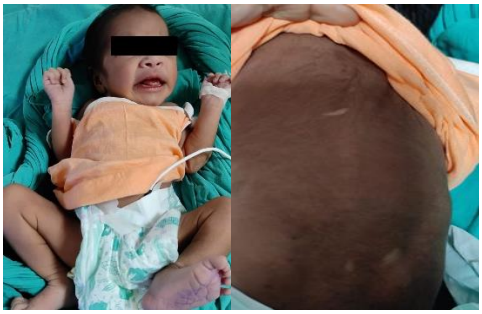
Prevalenceof Tuberous Sclerosis is 1 in 6000 to 1 in 10000 newborn. 2 foci for TSC:- TSC1 gene and TSC2 gene. TSC1 gene encodes a protein called Hamartin, TSC2 gene encodes a protein called Tuberin. Both TSC1 and TSC2 genes are considered as Tumor Supressor Genes.

The hallmark of TSC is involvement of the CNS.

Most common neurologic manifestationof TSC consists of Epilepsy, autism spectrum disorder, cognitive impairment.

In this report, we present a newborn infant with Rhabdomyoma of heart, Tuberous Sclerosis, who developed Respiratory distress few hours after birth.

A diagnosis of Tuberous Sclerosis was made as multiple skin lesions were presenton the whole body.



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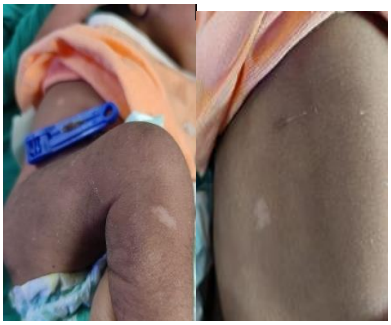

II. CASE DESCRIPTION :

FTNVD with AGA on day of life one was referred to our hospital, New Civil Hospital, Surat from sub-district hospital, Chikhli, Navsari for respiratory distress. Baby was born on 30/11/2021 at 3:30 am, FCH, Birth weight was 2.377kg, delivered by normal vaginal delivery. On examination: pulse-142/min, RR-64/min, activity-normal, O2 saturation-98% , RBS-94mg/dl, RS-AE=BS, CVS-S1&S2 Normal, CNS-NAD, P/A-soft.

Local Examination:-Multiple Hypopigmented patches were present all over the body.

The mother was 23yr old G1P1L1A0, history of consanguineous marriage was present, ANC USG (20-30 wks) showed mild polyhydramnios along with multiple rhabdomyomain the heart, Tuberous Sclerosis, besides this there was no other significant antenatal history.

Baby cried immediately after birth, no resuscitative measures were required. Baby was kept on O2 hood at 15 l/min for respiratory distress which developed a few hours after birth. Routine investigations were sent. Chest-X-ray showed presence of cardiomegaly. USG abdomen-pelvis & skull was normal. MBG, BBG- A+ve, Hemogram-Normal, CRP-2, S.Ca^{2+}-9.3, baby was kept on ryles tube feedsfor one day followed byspoon feeds and breast feeding.

2D-Echo showed multiple homogenous intracardiac mass suggestive of rhabdomyoma (3×4mm, 2×3mm, 4.5×5mm on LV side of intraventricular septum 27×15mm occupied RV apex). No outlet inlet obstruction. No pulmonary arterial hypertension, good Bi-ventricular function.

Rhabdomyoma likely to regress with age and risk of sudden cardiac death due to arrhythmia explained to relative adviced to follow-up after 6 months. Skin reference was taken for the hypopigmented patch.

Ophthalmology reference was taken in view of any retinal lesions but no abnormal retinal findings were noted.

Neurophysicianopinion taken, adviced for head circumference monitoring andMRI Brain to look for sub-ependymal nodule but relative gave negative consent for MRI Brain.

Patients relative was counselled regarding the disease in their own language of understanding (Gujrati) and also was informed about the poor general condition,poor prognosis explained, patient didn't want to continue hospitalization and hence discharge on request given, patient was discharged on supplement. Explained the possible risks child might have in future, also explained when to follow-up. Risk like cognitive behavioral, psychiatric and academic impairment, CCF, arrhythmias, lymphangioleiomyomatosis etc.

III. DISCUSSION :

Definite Tuberous Sclerosis diagnosis made:-atleast 2 major or 1 major + 2 minor features present. In addition pathogenic mutation in TSC1 or TSC2 gene is sufficient for diagnosis of Tuberous Sclerosis.

Major features of Tuberous Sclerosis complex:

1. Cortical dysplasias (including Tubers with cerebral white matter migration line).
2. Subependymal nodules.
3. Subependymal giant cell astrocytoma.
4. Facial angiofibromas (≥3) or forehead plagues.
5. Ungula fibromas (≥2).
6. Hypomelanotic macules (≥3, ≥5mm in diameter).
7. Shargreen patch.
8. Multiple retinal nodular hamartomas.
9. Cardiac rhabdomyoma.
10. Renal angiomyolipoma.
11. Pulmonary lymphangioleiomyomatosis.

Minor features of Tuberous Sclerosis complex :

1. Dental enamel pits (>3).
2. Intra-oral fibromas (≥2).
3. Retinal achromic patch.

4. Confetti skin lesions.
5. Non-renal hamartomas.
6. Multiple renal cysts.

Cardiac rhabdomyomas can be easily diagnosed by ANC USG. It is difficult to diagnose hamartomas in brain, kidney, heart and skin. In our case 2 major criteria were fulfilled hence we could easily make a diagnosis of Tuberous Sclerosis.

Rhabdomyoma are likely to regress with age but chances of sudden death due to arrhythmia was explained to the patients relative.

Need for Brain MRI and Renal USG every 1-3 yr, CT-Scan every 5-10 yr, Dental examination twice in a year, Ophthalmology and skin examination yearly, was explained to the patients relative also the relative was counselled to watch for signs/symptoms of raised intracranial pressure.

IV. CONCLUSION :

ANC USG is an important diagnostic tool for Tuberous Sclerosis, detected by fetal cardiac rhabdomyoma. In case if missed by ANC USG one should be suspicious if any hypomelanocytic patch is present on skin and further should be evaluated by a 2-D Echo for cardiac rhabdomyoma.

Pediatrician should be aware of prenatal and postnatal USG findings regarding Tuberous Sclerosis.

One should also be vigilant about the cardiac, neurological and renal findings on USG.

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