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A CASE REPORT OF MAPLE SYRUP URINE DISEASE

Pediatrics

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KEYWORDS

INTRODUCTION:

- Maple Syrup Urine Disease (MSUD) is a rare Autosomal Recessive inborn errors of metabolism of branched chain amino acids. MSUD is caused by deficiency of branched chain alpha ketoacid dehydrogenase enzyme complex.
- Caused by Mutations in genes for E1 Alpha, E 1 beta,E2 subunits
- Three types of MSUD -1.Classic 2.Intermediate 3.Intermittent
- Prevalence is estimated at 1:170000 births.

CASE REPORT:

14 months old child born to non-consanguinous couple 4th in birth order brought by parents with complaints of Global Developmental delay, feeding difficulties ,seizures since 4 days of life and abnormal smell of urine.

Antenatal/Natal history: Normal.

Postnatal history: Admitted in NICU for 21 days on 4th day of life got treated for neonatal seizures.

Family History: Similar complaints in maternal brother.
Anthropometry: Weight:10kg
Length: 76 cm
Head circumference: 45 cm

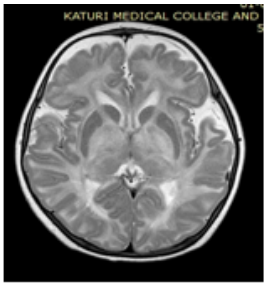
Head to Toe Examination: Normal
AF: 1.5X1.5 and patent.
Systemic examination: Hypotonia of all limbs
Plantar Bilateral extensors
Fundus: Normal

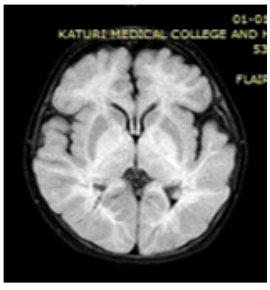
Investigations:
ABG: Metabolic Acidosis
Hemogram ,LFT, RFT,Serum Electrolytes:- Normal

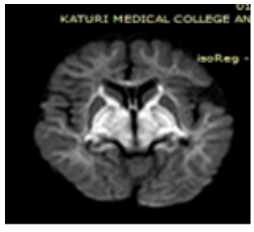
MRI: Diffuse hyperintense bilateral fronto-parieto-occipital white matter,thalami, brain stem and cerebellar dentate sulci noted

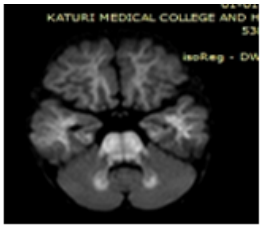
DW1 & ADC restrictions noted in bilateral peri rollandic white matter,thalami,internal capsule, brain stem and cerebellar dentate nuclei noted

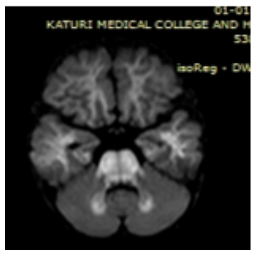
Prominent VR spaces noted
IMPRESSION: 1.Maple syrup urine disease (intermediate form)

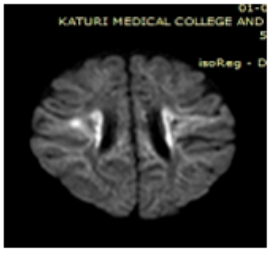












Name	:	Master D. KARITHIK			Collected	:	27/2/2017 11:30:00AM
Lab No.	:	236986013	Age: 1 Year	Gender: Male	Received	:	27/2/2017 2:14:00PM
A/c Status	:	P	Ref By :	Dr.PRAVEEN	Reported	:	4/3/2017 5:55:04PM
					Report Status	:	Final

Urine test (DNPH): positive.
Amino acid Quantitative Test:

Test amino acid	Result	Normal value
Leucine	185.53 Umol/L	47-155 Umol/L
Isoleucine	127.52 Umol/L	31-86 Umol/L
Valine	357.78 Umol/L	64-294 Umol/L

Impression-
Leucine, Isoleucine and Valine raised.
Suggesting MSUD.

DISCUSSION:

Intermediate MSUD: The child was diagnosed as intermediate type of MSUD based on typical MRI findings and confirmed the diagnosis by enzyme analysis.

Treatment: Child was treated with a trail of Thiamine and advised Diet low in BCAA. As there is limited availability of specialised formulas.

Prognosis: Long term prognosis of affected children remains guarded. Death may occur during any stressful event like surgery or in infection[(2)].

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