A RARE CASE OF MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOPATHY

ABSTRACT :

Mitochondrial Neuro Gastrointestinal Encephalopathy (MNGIE) is rare genetic disorer. It is characteristic by progressive gastrointestinal
dysmotility, cachexia, opthalmoplegia and leucoencephalopathy.We hereby report a case of MNGIE in a female.

KEYWORDS : Mitochondrial Neuro Gastrointestinal Encephalopathy.
Leucoencephalopathy

INTRODUCTION:

It mainly affects the Gastrointestinal tract and central nervous system. Clinical features starts at the age of 20 years and progressive in nature [1] . Dysmotility of gastrointestinal tract seen in majority of cases in MNGIE ,is a condition in which there is a difficulty in passage of food due to inactivation of muscles and nerves leads to early fullness, difficulty in swallowing, heartburns, nausea, vomiting after eating , pain in the abdomen , bloating and loose stools.

Generally patient diagnosed with MNGIE can have loss of appetite ,loose weight , ptosis,ophthalmoplegia , auditory impairment, Tingling, paresthesia,numbness,weakness of lower limbs . In MNGIE there is depletion of white matter of brain tissue which is the classical features of this syndrome,mostly less common (Leukoencephalopathy),Due to the autosomal recessive inheritance of TYMP gene in MNGIE which is a essential gene for maintaining proper level of mitochondrial thymidine .[1][2]

Blood levels of thymidine and deoxyuridine is raised in this syndrome and its diagnosed by identification of variations in *TYMP* gene .symptomatic management is the choice of treatment which includes maintaining the airway ,teaching new swallowing techniques, focal management for nausea vomiting and neuropathic features . further management incorporate diet support, antibiotics for bacterial infection in intestine ,aided education and and physical therapy like exertion ,exercise .[2] following medication chloramphenicol, tetracycline linezolid, ,valproate, phenytoin, and aminoglycosides should be avoided due to hindrance of mitochondrial function .[3]

CASE REPORT :

12 years female child of 3rd degree consanguineous parents came with complaints of abdominal discomfort, slurred speech, hearing disturbance, loss of weight for 7 years.
On examination: Child had opthalmoplegia, ptosis, B/L mixed hearing loss, slurred speech and cachexia.(Fig-1,2)
Per abdomen examination: Visible gastric peristalsis seen.(Fig-3)

INVESTIGATION:
USG abdomen shows hypoplastic uterus.
MRI shows white matter leukodystrophy.(Fig-5)
X-ray abdomen shows pseudo-obstruction.(Fig-4)
Hormonal assay shows elevatedLH and FSH, serum lactate and CSF lactate pyruvate is increased.
Muscle biopsies: ragged red fibre in gomoritrichrome stain.

DISCUSSION
Diagnosis of MNGIE is made in the child by the presence of h/o consanguinity, clinical findings and neuroimaging (4).
CSF protein and lactate are raised andmuscle biopsies: ragged red fibre in gomoritrichrome stain (5).
Muscle biopsies: ragged red fibre in gomoritrichrome stain.
Molecular genetic testing for routine TYMP gene mutation has been sent, reports awaited (6).

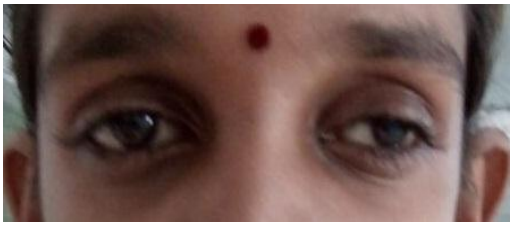

Fig 1: A 12-year-old female patient -opthalmoplegia, ptosis

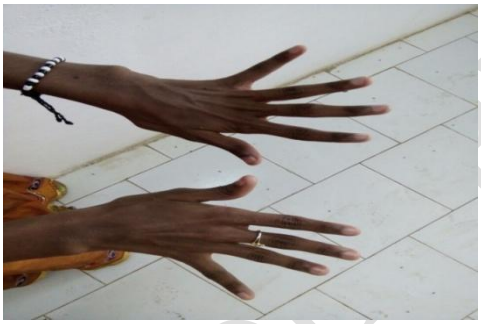

Fig 2:cachexia.

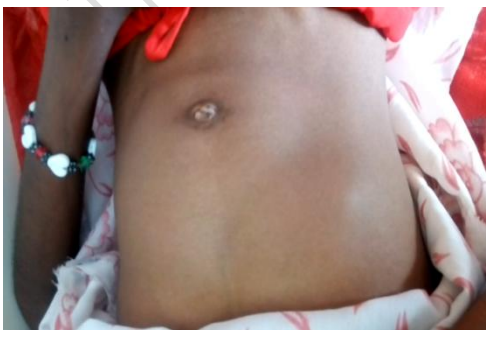

Fig 3: Visible gastric peristalsis

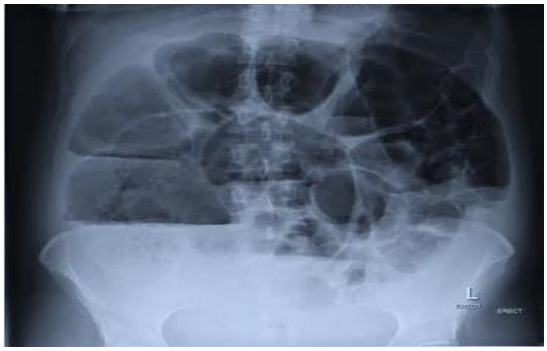

Fig 4: pseudo-obstruction

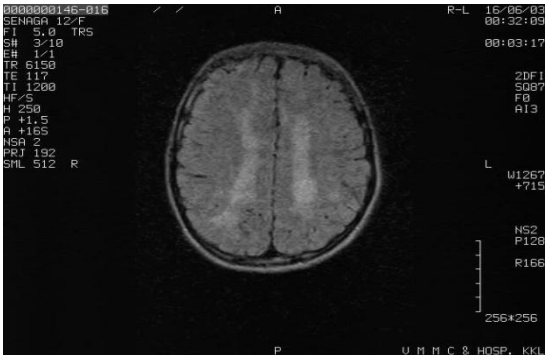

Fig 5: white matter leukodystrophy

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