Abstract:

Acute intermittent porphyria is an unusual presentation with median arcuate ligament, both of these two conditions may mimic each other in their symptomatology and may confuse physician to pinpoint the diagnosis.A 21 yrs old lady who presented with acute pain abdomen and low back ache. On evaluation, a condition called median arcuate ligament was detected radiologically along with the condition of acute intermittent porphyria by testing positive for urine for porphobilinogen.Since the occurrence of these two conditions is rare together and symptomatolgy also almost similar and cofusing to mislead a physician,so thought to report the case.

Introduction:

Acute intermittent porphyria (AIP) is a rare hereditary disorder of haemoglobin synthesis characterised by episodes of severe abdominal pain and passage of excess amount of porphobilinogen(PBG)in the urine.Abdominal pain in AIP may stimulate acute surgical abdomen leading to an unwanted laparotomy.Rarely,generalised epileptic fits and neuropsychiatric disturbances may occur.It is an autosomal dominant condition with incomplete penetrance and potentially very distressing and agonising for patients with its recurrent attacks of symptoms.The key point in the management is to suspect and confirm its diagnosis as early as possible so that to avoid inappropriate treatments which may exacerbate the crisis. But in our case the confusion arose when we found an underlying coexisting condition called Median Arcuate Ligament Syndrome.

Median Arcuate Ligament Syndrome(MALS,also known as celiac artery compression syndrome,Dunbar syndrome)is a condition characterised by abdominal pain attributed to compression of celiac artery and possibly the celiac ganglia by the median arcuate ligament.Unlike the porphyric pain,MALS presents with the symptomps of postprandial pain abdomen,nausea or anorexia ,weight loss and abdominal bruit at times.Median arcuate ligament is a ligament formed at the base of the diaphragm where left and right diaphragmatic crura join near the 12^{th} thoracic vertebra.This fibrous arch sometimes in one quarter of normal individuals comes in contact with the celiac artery or celiac ganglia compressing it producing the syndrome.The mainstay of treatment in MALS is surgical approach to divide or separate the median arcuate ligament to relieve the compression along with removal of celiac ganglia or angioplasty and stenting.

We report here this case due to its rarity and also because of its cofusing picture in the presence of Median Arcuate Ligment Syndrome(MALS)which also mimics

almost the similar signs and symptoms like that of porphyria.

Case Report:

A 21 years female,a graduate student presented with the complaints of low back ache and severe pain abdomen from 14 days duration.The pain was progressive and refractory to many analgesics she used to have over the counter.Her back pain was confined to low back around lumbosacral region along with diffuse abdominal pain.There is no significant past history relevant to her pain nor any history of loss of appetite , weight loss and postprandial nausea or exaggeration of abdominal pain.Her general examination was normal with normal vitals.

On examination,her abdomen was soft,nontender with good peristaltic sounds without any abdominal bruit.there was no local spinal tenderness or deformity.

Investigation:

Routine blood investigations-

TLC:8500/cumm,RBC:4.6lacs,HB:9.6,PLATELET:222,PCV:30.7,MCV:66.7,MCH:20.7,MCHC:31. 1,Na:131 ,K

:3.37,BUN:8.0,CREATININE:0.68,EGFR:116.09,CPK:540,S.AMYLASE:53,S.LIPASE:117,RBS:12 8.Mg:2

T.BILIRUBIN:1.0,T.PROTEIN:6.3,GLOBULIN:3.0,ALBUMIN:3.3,A/G:1.1,SGOT:48,SGPT:106,GGT:

24,ALP:106,PERIPHERAL BLOOD SMEAR:moderate anisocytosis,normocytic normochromic to microcytic hypochromic with normal WBC,platelets wth absence of any hemoparasites or abnormal cells.

URINE FOR PORPHOBILINOGEN(URINE,WATSON,SCHWARTZ METHOD):PRESENT.

serum osmolarity:246.97,urine osmolarity:334.21,urine Na:146,TSH:1.47 ECG:normal sinus rhythm,Chest xray: normal.VIRAL SEROLOGY:nonreactive.

MRI SPINE: loss of lumbar lordosis with reduction of L4-L5 with mild central bulge at L5-S1.

CECT ABDOMEN: Hooking of celiac artery in the ostioproximal segment with mild focal stenosis likely of median arcuate ligament syndrome with element of constipation,bulky ovaries suggestive of PCOD.

UPPER GI ENDOSCOPY: Normal study. CT ANGIOGRAPHY OF ABDOMEN:

Diagnosis:

On the basis of history,clinical profile and laboratory reports,the patient is diagonised to be a case of ACUTE INTERMITTENT PORPHYRIA(AIP).

She was treated conservatively for her excruciating pain and vomiting and nausea with inj.tramadol 50 mg iv Thrice daily with ondansetron,tab olanzepam O.25mg twice daily,tab Gabapentin 300mg 8 hrly,25% iv dextrose(300-500gm/day) and high carbohydrate diet and also she had to be treated in intensive care unit with high dose fentanyl iv infusion followed by later on by fentanyl patch.

Discussion

In our patient,the diagnosis of porphyria was made on the basis of clinical manifestations and presence of PBG in urine.It was measured qualitatively by Watson and Schwatz method,although not a confirmatory test but quantitative measurement of PBG is not easily available in our country.

Also,in our case it was an incidental finding that there was evidence of presence of MEDIAN ARCUATE LIGAMENT SYNDROME(MALS) radiologically while evaluating for intractable pain abdomen.Though the clinical profile of median arcuate ligament syndrome also mimics the presentations of AIP but there were few points those go against the diagnosis of MALS.In our case,there was no weight loss or postprandial abdominal pain which is common in MALS.Also,the patient in our case improved and became pain free only with the treatment of opioids analgesics and high glucose therapy/carbohydrate diet which cannot be possible in case of MALS as because the definitive therapy of MALS is surgery with division of median arcuate ligament to release the celiac ganglion and celiac trunk from the hooking.

Review of Literature:

Acute porphyrias are characteristically hepatic porphyria,presenting with neurovisceral and psychiatric disturbances like abdominal pain(90%cases),constipation,insomnia,depression,hallucinations .in acute porphyric

crisis,encephalopathy varying from confusion to frank psychosis can occur concomitantly with hypothalamic involvement and metabolic derangement of inappropriate secretions of ADH.generalised seizures,myoclonus or coma may be observed due to neurological effects or hyponatraemia.

Polyneuropathy and painful flaccid paralysis predominantly involving upper limbs:preferentially affecting proximal musculature with occasional sensory involvement.motor weakness may be asymmetric or focal.cranial nerves may get affected.progressive muscle weakness can lead to life threatening respiratoty and bulbar paralysis.

Autonomic disturbances may manifests as urinary retention,paralytic ileus,restlessness,trmors,excessive sweating,tachycardia,fluctuating blood pressure.sometimes profound hypotension requiring inotropes then bradycardia and sudden cardiacarrest.

The reason for neurological involvement is poorly understood.direct neurotoxicity of delta ALA by interaction with GABA receptor,altered tryptophan metabolism or a neural respiratory heam dependant enzymatic deficiency in nerve cells has been hypothesised.

Diagnosis of aip requires a clinical suspicion with documentation of ALA and PBG in fresly voided urine.classic burgundy red discoloration of lond stored urine or Watson –schwartz test usig Ehrlich,s aldehyde reagent are useful for screening.quantitative measuresment of of PBG and ALA in urine or erythrocyte hydroxymethylbilane synthase test are more confirmatory and reliable..but exorbitant costs limits their use.

Patientswith aip may require ventilator support for progressive ascending paralysis with respiratory and or bulbar involvement.someties so ai may mimic GBS.absence of lymphocytosis with raised csf protein favors its diagnosis of GBS rather than AIP.varying neuropsychiatric manifestations will require treatmentwith analgesics ,antiepileptics and sedatives.but its alws better to avoid drugs which are metabolised through liver .drugs like barbiturates, phenytoin,valproate,carbamezapine always unsafe and avoided in aip.propofol ,gabapeentin,magnesium sulphate can be used in aip.

Haem in the form of haematin,heam albumin or heam arginate 3-4 mg/kg/day infused daily for3 to 4 days is the treatment of choice in aip.haem acts by repressing the ALA synthase enzyme,further suppressing the production of haem precursors.but associated high cost and unavailability is the drawback.alternatively carbohydrate rich food or intravenous dextrose in higher doses(300-500gm/day)blocks induction of the enzyme.In future Allogenic liver

transplantation and liver directed gene therapy is coming up as the last resort of treatment in patients with recurrent attacks but these options are still under research and debatable.

To conclude,aip is a special group of patints who need special care.high clinical suspicion,early diagnosis and management of ai can prevent the patient to go through many unwanted treatment and intervention and morbidities.aip should alws be suspected in a patint with neuropsychiatric manifestions with gastrointestinal symptoms even the family members also should be screened for the same.

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