An Interesting case of Peripheral neuropathy

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GUIDE –
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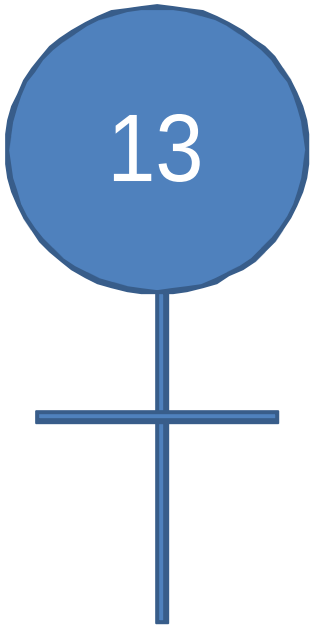
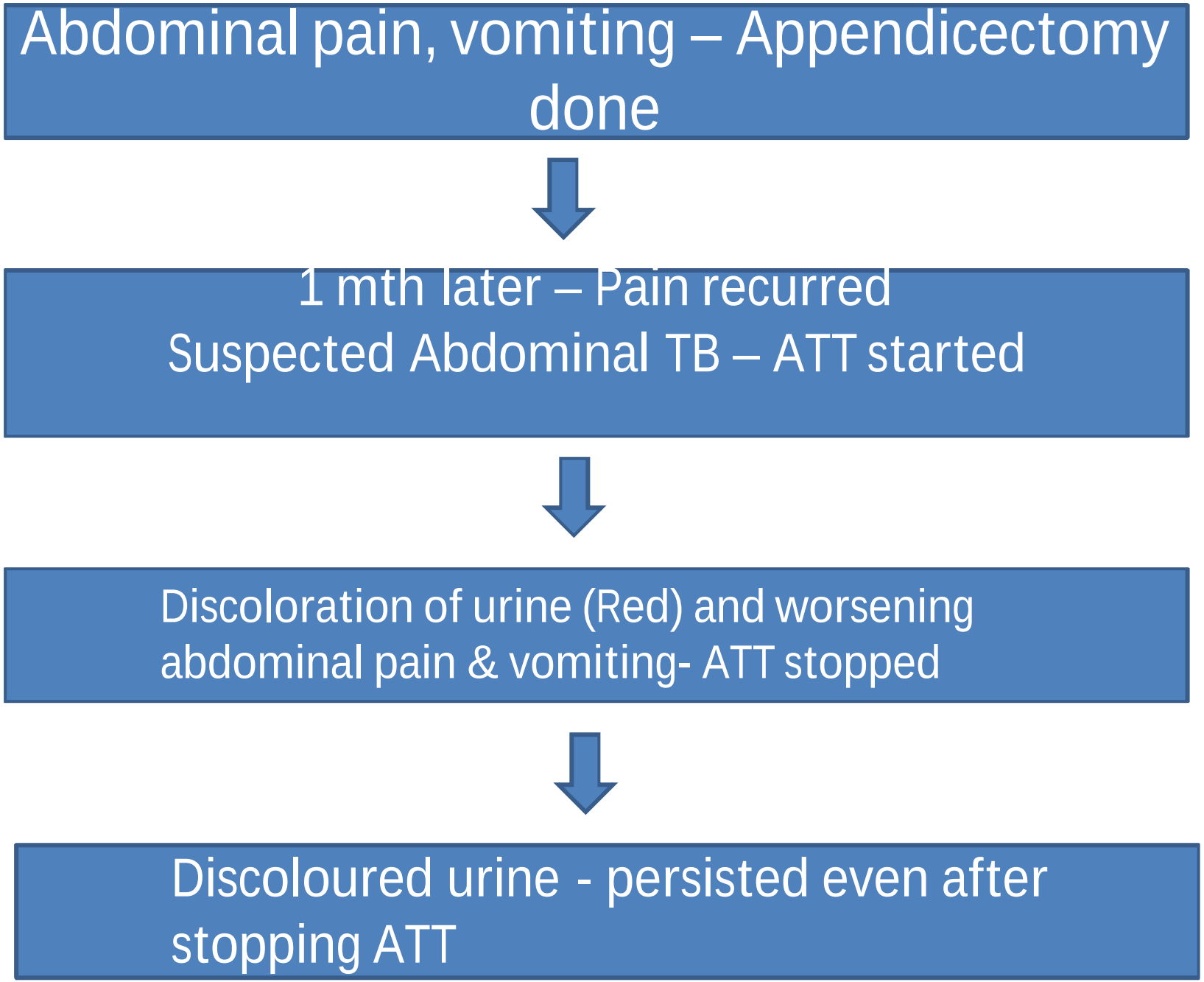
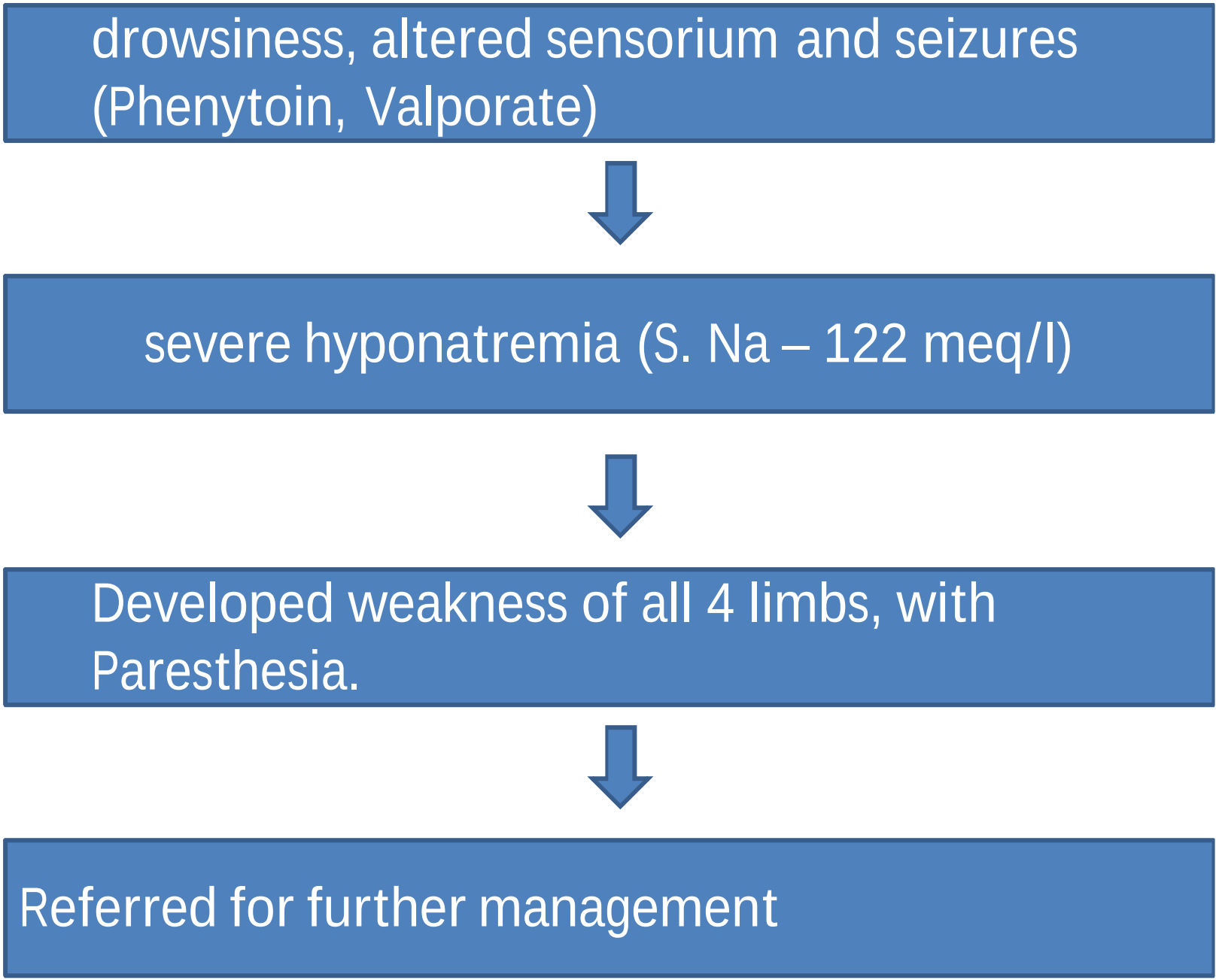
HISTORY



- ❖ Abdominal pain on and off for past 6 months
- ❖ Vomiting for past 6 months
- ❖ Drowsiness – 1 week


Abdominal pain, vomiting – Appendicectomy done
1 mth later – Pain recurred
Suspected Abdominal TB – ATT started
Discoloration of urine (Red) and worsening abdominal pain & vomiting- ATT stopped
Discoloured urine - persisted even after stopping ATT


drowsiness, altered sensorium and seizures (Phenytoin, Valporate)
severe hyponatremia (S. Na – 122 meq/l)
Developed weakness of all 4 limbs, with Paresthesia.
Referred for further management

EXAMINATION

Lethargic

GCS – 14/15

Moaning with abdominal pain

Some dehydration (+)

No Meningeal Signs

Vitals- stable

Systems- normal

PROVISONAL DIAGNOSIS

Recurrent abdominal pain

vomiting

unexplained hyponatremia

urinary discoluration

? Acute Intermittent Porphyria + SIADH

COURSE IN THE HOSPITAL

IN PICU:

- Dehydration - 5% correction given (Normal Saline)
  - Maintenance fluid– 10% dextrose with NS
- baseline investigations -

INVESTIGATIONS	RESULT
S. ELECTROLYTES	SODIUM 129
LFT, CBC, URINE R/E	NORMAL
SERUM OSMOLALITY	265
URINE OSMOLALITY	847
Urine Na	> 95

  - Suggestive of SIADH
  - 3% Nacl Correction started

- Seizures - Phenytoin & Valporate stopped
  - Leviteracetam started (safe drug if AIP)

- Urinary discoloration & Suspected AIP
  - Urine routine & CPK – Normal
  - 24 hour urinary porphobilinogen

INVESTIGATION	RESULT
U.PHORBILINOGEN	NEGATIVE
USG ABDOMEN	NORMAL
NERVE CONDUCTION STUDY	PERIPHERAL NEUROPATHY
CPK	NORMAL

Porphyria Screening

Sample : 24 Hr Urine
24 Hr Urine Volume : 1.4 L
Urine Creat : 0.28 g/ L

	Metabolites	Observed Values	Reference Range
1	Delta Aminolevulinic Acid (ALA)	218 uMol/24 hr	< 49 uMol /24 hr
	Delta Aminolevulinic Acid (ALA)	556 uMol/g Cr	< 56 uMol/g Cr
2	Porphbilinogen (PBG)	224 uMol/24 hr	< 7.5 uMol/24 hr
	Porphbilinogen (PBG)	571 uMol/g Cr	< 12.3 uMol/g Cr
3	Total Porphyrin	1,082.99 ugm/24 Hr	< 150 ugm/24 Hr
	Total Porphyrin	3,867.84 ugm/g Cr	< 175 ugm/g Creat.

Impression: Grossly elevated ALA, PBG and Total Porphyrin, suggestive of Acute Porphyria (AIP / VP / HCP).

Urine Porphyria Screening

	PATIENT INITIAL SAMPLE
TOTAL PORPHYRIN/CREATININE	4,934.96 (<174 ug/g creat.)
Delta Aminolevulinic Acid (ALA)	218 uMol/24 hr (< 49 uMol /24 hr)
Porphbilinogen (PBG)	224 uMol/24 hr (< 7.5 uMol/24 hr)
Total Porphyrin	1,082.99ugm/24 Hr (< 150 ugm/24 Hr)

IMPRESSION :Total coproporphyrin along with grossly elevated Uro / Copro porphyrin ratio, suggestive of AIP.

Blood Porphyria Screening by HPLC

Metabolites	Observed Values	Reference Range
Free Protoporphyrin	38.02nMol/L	9.0 – 89.0 nMol/L
Free Protoporphyrin – Relative amount	7.58 %	< 30 %
Zinc Protoporphyrin	28.78uMol/mol Hb	< 40 uMol/mol Hb
Zinc Protoporphyrin – Relative amount	92.42 %	> 70 %
Total Protoporphyrin	501.68nMol/L	< 540 nMol/L

Impression: Normal
Protoporphyrin in blood

Porphobilinogen Deaminase Enzyme Activity

ENZYME ACTIVITY HEPARINIZED BLOOD	OBSERVED VALUES	REFERANCE RANGE
patient	16.32 (70.31 %)	Normal :18.5 – 23.21 nmol/Lit/s
Normal Control (Negative Control)	23.21	
AIP Control Sample (Positive Control)	6.65 (28.65%)	AIP carriers : 6 – 10 nmol/Lit/s

IMPRESSION :Low PBG Deaminase enzyme activity.

Urine Porphyria Screening by HPLC

Sample : 24 Hr Urine
24 Hr Urine Volume : 1.4 L
Urine Creat : 0.28 g/L

	Metabolites	Observed Values	Reference Range
1	Uro porphyrin		< 27 ug/24 hrs
	Uro porphyrin / Creatinine	4,358.39	< 33 ug/g creat.
2	**Uro Porphyrin Isomers**		
	Uro Porphyrin I	45.81 %	53 – 79 %
	Uro Porphyrin III	54.19 %	21 – 47 %
	Hepta Porphyrin	-	< 8 ug/ 24 hr
	Hepta Porphyrin / Creat	22.32	< 10 ug/g creat.
	Hexa Porphyrin	47.15 (ug/g cr)	< 6 ug/24 hr
	Penta Porphyrin	-	< 4 ug/24 hr
	Penta Porphyrin / Creat	38.72	< 5 ug/g creat.
3	**Copro Porphyrin**		
	Copro porphyrin Total		< 100 ug/24 hrs
	Copro porphyrin Total/ Cr	468.38	< 120 ug/g creat.
	Copro porphyrin I	21.77 %	17 – 31 %
	Copro porphyrin III	78.23 %	69 – 83%
4	**Uro / Copro porphyrin**	**9.31**	**0.07 – 0.65**
5	**Total Porphyrin**		**< 145 ug/24 hrs**
	Porphyrin / Creat Ratio	**4,934.96**	**<174 ug/g creat.**

Impression: Grossly elevated Uro porphyrin (Uro III > Uro I), mildly elevated hepta-, hexa-, and penta- coproporphyrins, Total coproporphyrin along with grossly elevated Uro / Copro porphyrin ratio, suggestive of AIP.

- Carrier screening will be needed for parents
- Mother found to be a carrier.
- Mother and child counselled for safe and unsafe drugs

Child is on regular follow up and doing well(on High carbohydrate diet)

DISCUSSION

- Neuropsychiatric manifestations, SIADH, discoloured urine – Suspect Acute Intermittent Porphyria
- Proving AIP is difficult
  - needs multiple sampling or 24 hours urine collection
  - HPLC is the preferred method for diagnosis
  - Investigations are expensive
- Haemin Therapy is expensive and will decrease duration of illness only in acute phases.
- Good response with high dose of dextrose.
  - Use along with NS (to avoid hyponatremia)

LITERATURE REVIEW

Indian Journal of Psychiatry
Official Publication of the Indian Psychiatric Society
Home
Current issue
Instructions
Submit article

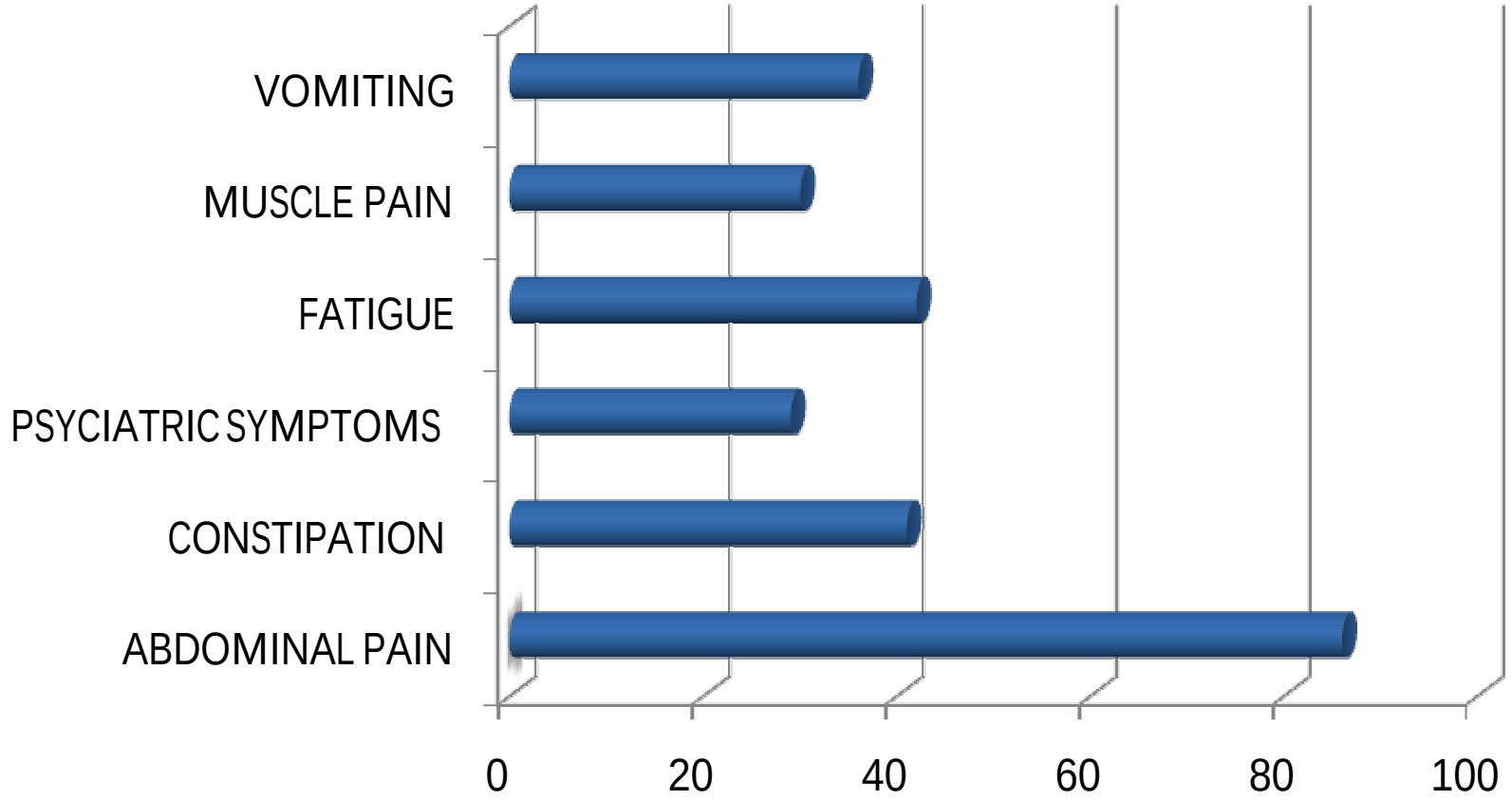
Indian J Psychiatry. 2006 Jul-Sep; 48(3): 189–192.
doi: 10.4103/0019-5545.31584
PMCID: PMC2932991

An analysis of six cases of acute intermittent porphyria (AIP)

Soumitra Ghosh,* Pranit KR. Chaudhury,** and Hiranya K. Goswami***

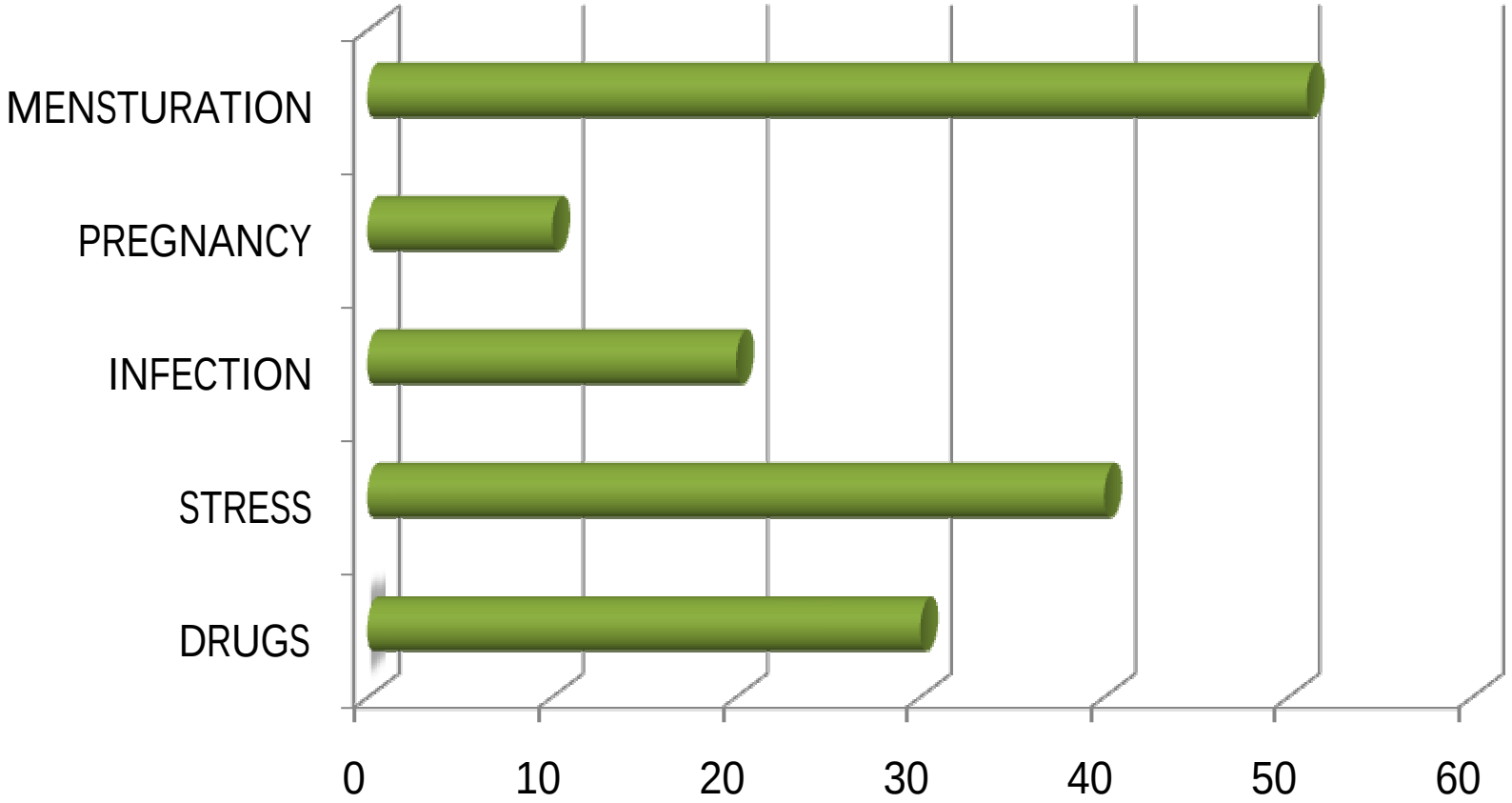
- 6 patients (5 women, 1 man; mean age 28.5 years) of (AIP), presented to the Psychiatry OPD over a period of one year.
- Among the 6 patients, 4 had abdominal pain, 5 had autonomic instability, all 6 had mental symptoms, 3 had depression, 2 came in delirium, and 3 had an episode of seizure.

Clinical aspects of acute intermittent porphyria in northern Sweden: a population-based study



Frequency of symptoms (%) during acute attacks reported by 149 gene carriers with manifest AIP

Clinical aspects of acute intermittent porphyria in northern Sweden: a population-based study



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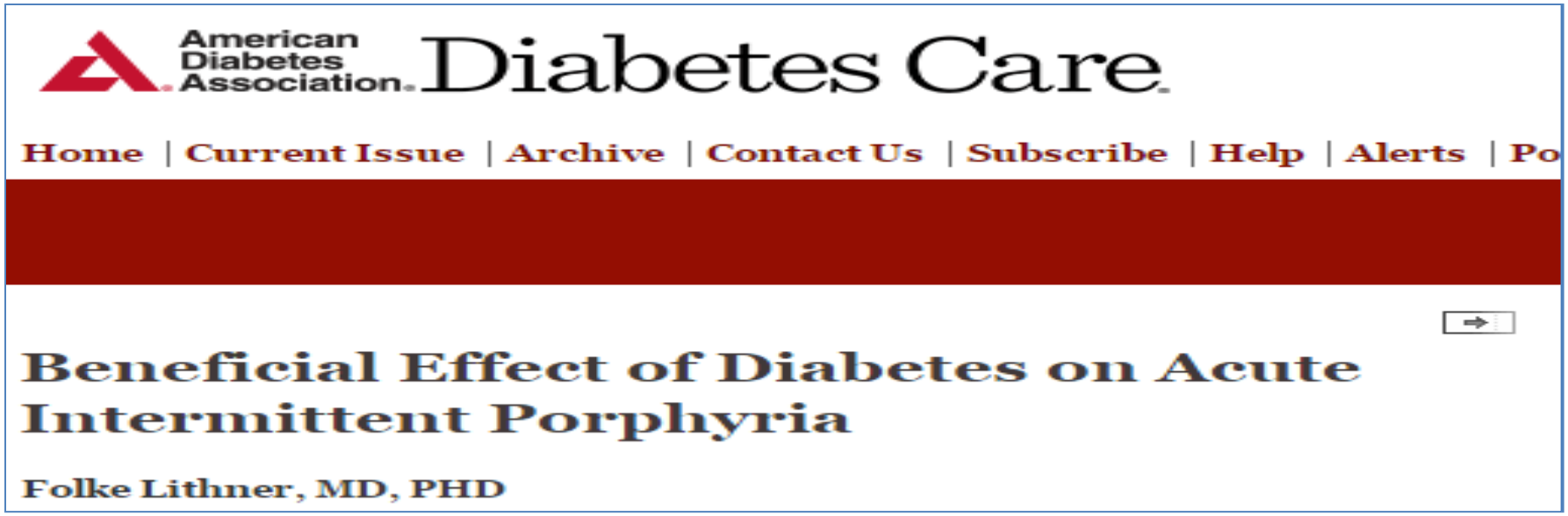
. Frequency of precipitating factors (%) to AIP attacks reported 149 gene carriers with manifest AIP

Journal, Indian Academy of Clinical Medicine
Vol. 3, No. 3 July-September 2002

- IV haeme is more effective than glucose in reducing porphyrin precursor excretion and probably leads to more rapid recovery.
- 3 to 4 mg/kg body weight in the form of haematin, haeme albumin, or haeme arginate may be infused daily for 4 days.
- The rate of recovery from acute attack depends on the degree of neuronal damage and may be rapid (1-2 days) with prompt therapy. The response to haeme therapy is unsatisfactory if treatment is delayed



- Recurrent AIP attacks ceased when the patients became diabetic.
- None of the 16 diabetic patients with AIP had HCC.
- Of all the 30 AIP patients with HCC registered, none had diabetes.
- This suggests that diabetes also counteracts HCC in AIP patients, probably by normalization of ALA

ACKNOWLEDGEMENT

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- NIRMAN, Mumbai

THANK YOU

PEADENDO 2015

Paediatric & Adolescent Endocrinology Conference for Postgraduates & Practitioners

Organised by:

Department of Paediatrics, SRMC & RI and ICH&HC

In association with IAP – TNSC & IAP - CCB

Date : 30/8/15 *Time: 8 am to 5 pm*

Venue: Hotel Ramada, Chennai

Program Highlights:

- Renowned National & International Speakers
- Booklet on "Treatment of Paediatric Endocrine disorders – A pocket guide"
- PG QUIZ: one team from each institution (2 PGs – M.D/DNB)
  - Preliminary rounds in the venue– top scoring 4 teams will go for final round

Applied for Tamilnadu Medical Council credit hours....

Registration: Rs. 700/-

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