A SPIKY PROBLEM

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- 7 Month old girl, 2nd born / 3rd degree consanguineous parents
- c/o not gaining adequate weight of her age
- H/o regurgitation of feeds present 1-3 episodes /day
- Stools 2-3 episodes/day
- No H/o / respiratory distress/
- No H/o feeding difficulties
- No H/o fever / RRTI/ ear discharge
- No H/o convulsions
- No H/o polyuria / bone changes

Birth and Developmental history

- Full term, Birth weight - 2.75Kg
- Neonatal uneventful
- Milestones: gross motor developmental delay
  - Social and language milestones were appropriate for age
- Diet : exclusive breastfeeding, 10 feeds/day
  - Position and attachment good

On Examination

- Child alert
- Pallor +
- Malnourished, Loss of buccal pad of fat
- Cardiovascular and respiratory system- normal
- Abdomen distended, no organomegaly
- Nervous system- deep tendon reflexes 1+, normal power

Anthropometry

- Weight - 2.670 kg ; Weight for age <-3 SD
- Length - 50 cm ; Length for age <-3 SD
- Weight for length <-3 SD
- Head circumference - 35.5cm
- Mid upper arm circumference - 7cm

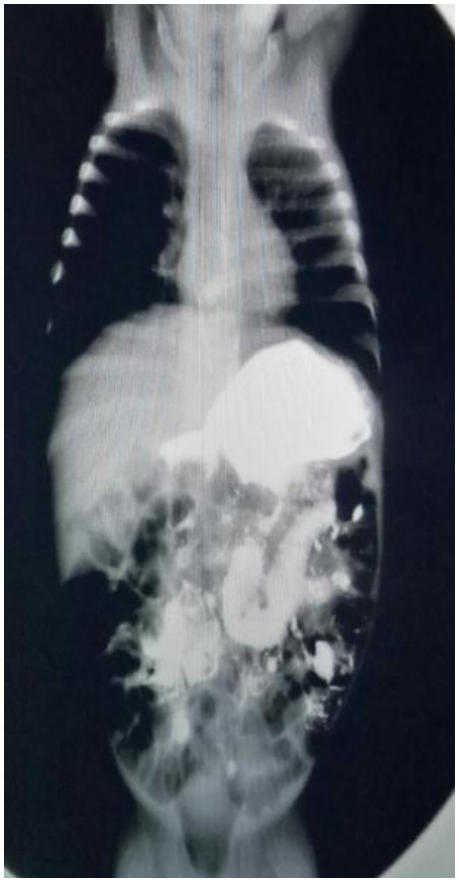
SAM

Investigations

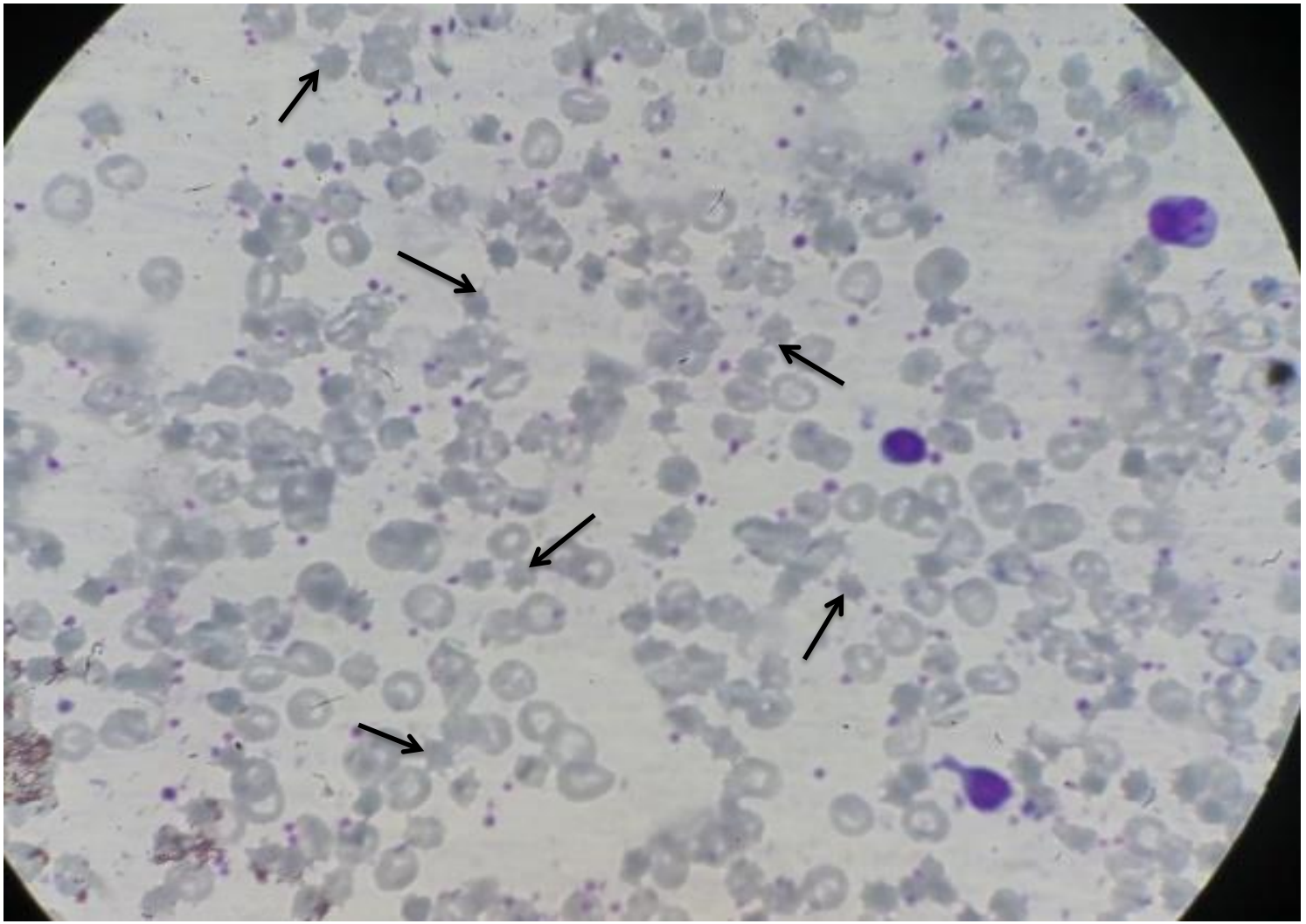
- Complete blood count - normal
- Renal function, liver function ,serum electrolytes -normal
- Serum proteins –normal
- USG abdomen - normal
- Sweat chloride -16 meq/l

Stool for fat globules -10 /hpf

Barium meal-malabsorption pattern



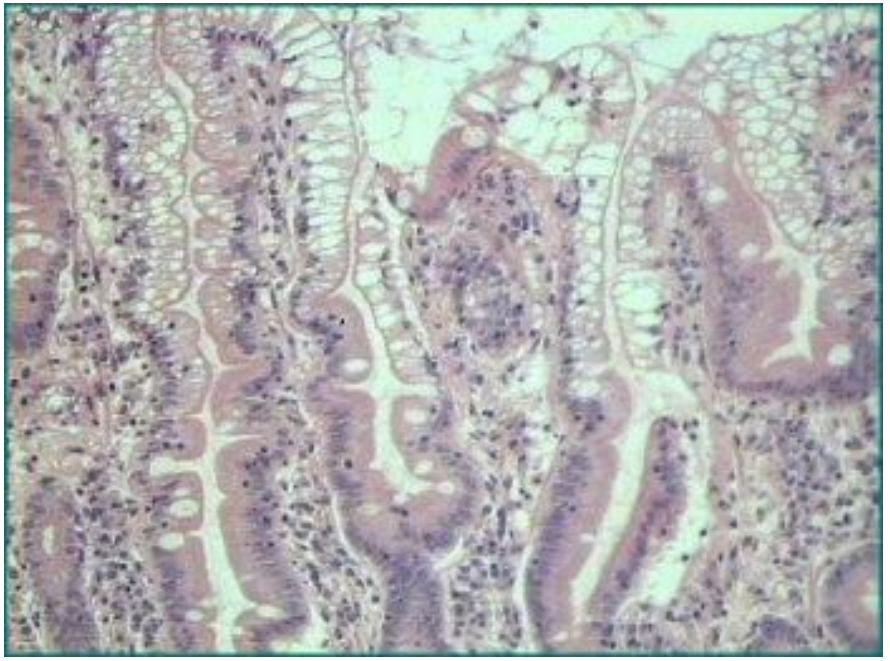
- Peripheral smear showed acanthocytes > 50 %



Baseline lipid profile

	Patient values	Reference values
S cholesterol	**40**	**very low (< 50 mg/dL)**
S triglycerides	10	<130
Hdl	40	>45
Vldl	2	2-30
Ldl	1	<130
Apoa1	106	110-160
Apo b	**6**	**40-125**

- Endoscopy showed pale duodenal mucosa
- Biopsy - preserved villous architecture with fat filled enterocytes suggestive of lipid transport disorder
- Fundus - normal



Final diagnosis

- Severe growth retardation
- Fat malabsorption
- Very low cholesterol, LDL and VLDL
- Markedly reduced ApoB
- >50% acanthocytes
- Small bowel biopsy – fat filled enterocytes with preserved villous architecture

HYPOBETALIPOPROTEINEMIA

Abetalipoproteinemia and Familial Hypobetalipoproteinemia (FHBL)

- Rare diseases - autosomal recessive ,characterized by fat malabsorption, spinocerebellar degeneration, acanthocytic red blood cells, and pigmented retinopathy.
- Improper packaging and secretion of apolipoprotein B-containing particles
- FHBL - caused by an autosomal, codominant mutation in the gene for apoB (APOB), which is carried on chromosome 2.
- Results in a truncated form of apoB
- Affected infants - appear normal at birth, but by the first month of life, develop steatorrhea, abdominal distention, and growth failure

- Vitamin E

  - Affected in 3 steps in the pathway of vitamin E absorption

1st pathway :

First, along with other fat soluble vitamins, the fat **malabsorption** decreases the absorption of vitamin E

2nd pathway

Second, the small amount of vitamin E that may be absorbed can not be efficiently secreted by the intestine because of the **defect in the chylomicron secretion**.

3rd pathway

Third, any vitamin E that is delivered to the liver also can not be secreted because of the **defect in the VLDL secretion.**

Treatment

- Management of FHBL and ABL essentially same
- Dietary manipulation - Severe restriction of long-chain fatty acids to 15 g per day . Improve the complications of fat malabsorption.
- In infants with failure to thrive, brief supplementation with medium-chain triglycerides may be necessary, but the amount must be closely monitored to avoid liver toxicity.

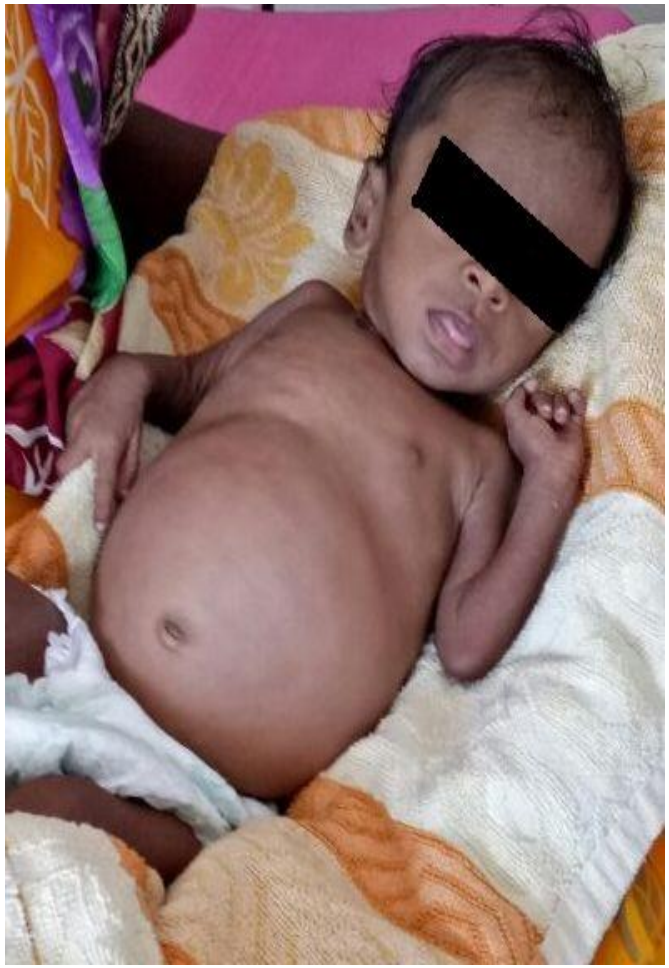
- Vitamin supplementation - Very large doses of oral vitamin E (100-300 mg/kg/d) are used to raise the tissue vitamin E concentration and to prevent neurologic complications in homozygotes.
- Vitamin A (10,000-25,000 IU/d) supplementation is instituted .
- Vitamin k 5mg

Prognosis

- Spinocerebellar degeneration secondary to vitamin E deficiency manifests in loss of deep tendon reflexes progressing to ataxia and lower extremity spasticity by adulthood
- Progressive pigmented retinopathy associated with decreased night and colour vision blindness
- Early treatment with supplemental vitamins especially vitamin E significantly slow down development of neurological sequelae

Before Vitamin Supplement

Two Weeks After dietary supplementation



Malabsorption + Acanthocytosis + hypocholesterolemia = Abetalipoproteinemia

THANK YOU