# An uncommon cause of anemia

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Unit 5

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- 17 year old male presented with c/c of:

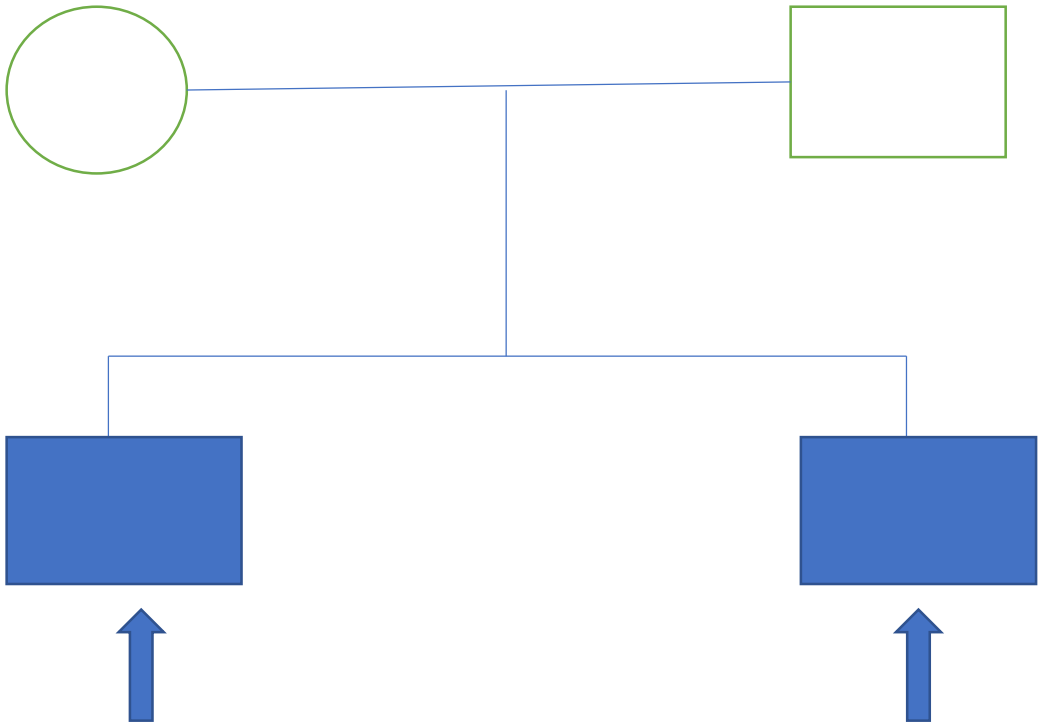
- Exertional breathlessness since 1 year
  - Insidious onset
  - Gradually progressive (NYHA 0 to NYHA 3)
  - No history of chest pain/ palpitations/ sweating/ swelling of lower limbs/ giddiness/ fever/ cough/ cold/ sore throat
  - No history of yellowish discolouration of eyes.
  - No history of malena/ hematochezia.

- On further enquiry patient also complains of :
- Night blindness
  - Since childhood
- Bilateral hearing loss
  - Since childhood
  - Non-progressive
  - No history of ear discharge, fever, tinnitus, earache, headache, giddiness

- PAST HISTORY: No previous hospital admissions.

- PERSONAL HISTORY:
- Mixed diet.
- Appetite normal.
- Normal sleep
- Normal bowel/ bladder
- No addictions
- Growth and developmental history normal since birth.

- FAMILY HISTORY:
- History night blindness and hearing loss in brother present.



- General examination:
- PR:  100 BPM
- BP: 100/60 mmHg
- RR: 22 /min
- SpO2: 98% on room air
- BMI: 20 Kg/m2

- Pallor present
- JVP: normal
- No oedema/ lymphadenopathy/ icterus/ clubbing/ cyanosis.
- No Bitot’s spots, corneal xerosis or hyperkeratotic patches.

- SYSTEMIC EXAMINATION:

- CNS: conscious, oriented to time, place and person.
  - Whispering test: decreased bilaterally
  - ABC: bilaterally decreased.

- CVS: S1S2 heard, normal. No murmurs appreciated.

- RS: Air entry bilaterally equal. No adventitious sounds heard.

- P/A: soft, non-tender. No organomegaly.

# PROVISIONAL DIAGNOSIS:

- Anaemia ?cause with Night blindness with bilateral sensory neural hearing loss.
- Due to:

1. Iron deficiency
2. Chronic malnutrition

| Hb | 2.5 |
|---|---|
| TLC | 7700 |
| PLT | 1,90,000 |
| MCV | 62.8 |
| BIL (T/D) | 0.60/ 0.23 |
| AST | 07 |
| ALT | 10 |
| ALP | 106 |

| PROTEINS(TOTAL/ ALBUMIN) | 6.10/ 3.70 |
|---|---|
| UREA | 268 |
| CREATININE | 18.95 |
| Na | 136 |
| K | 4.30 |
| Ca/ PO4 | 5.30/ 9.0 |
| URINE R/M | PROTEINS 3+ |
| UPCR | 7.78 |

| Sr IRON | 82 | VBG | |
|---|---|---|---|
| Sr FERRITIN | 230 | pH | 7.26 |
| Vit B12 | 451 | PCO2 | 19 |
| LDH | 540 | HCO3 | 8.30 |
| Sr ERYTHROPOETIN | 4 | | PRIMARY METABOLIC ACIDOSIS WITH COMPENSTARY RESPIRATORY ALKALOSIS |
| PBS | NORMOCYTIC, AND MILDLY HYPOCHROMIC. ANISOCYTOSIS+ | | |
| RETIC COUNT | 0.50 | CHEST X-RAY | NO ABNORMALITIES DETECTED |
| 25-OH Vit D | 15.20 | ECG | SINUS TACHYCARDIA |

- Skin biopsy:

  Inconclusive.

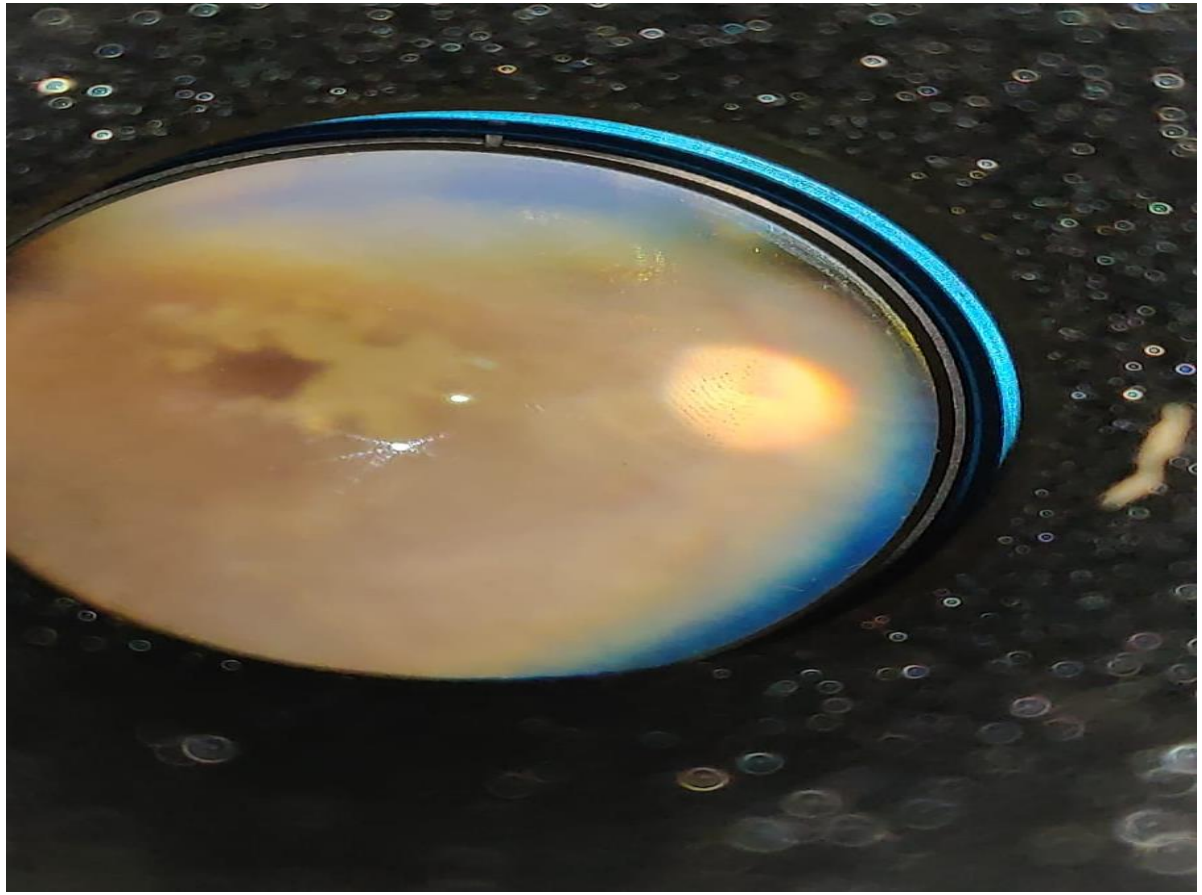
  Advice: DNA studies for confirmation.

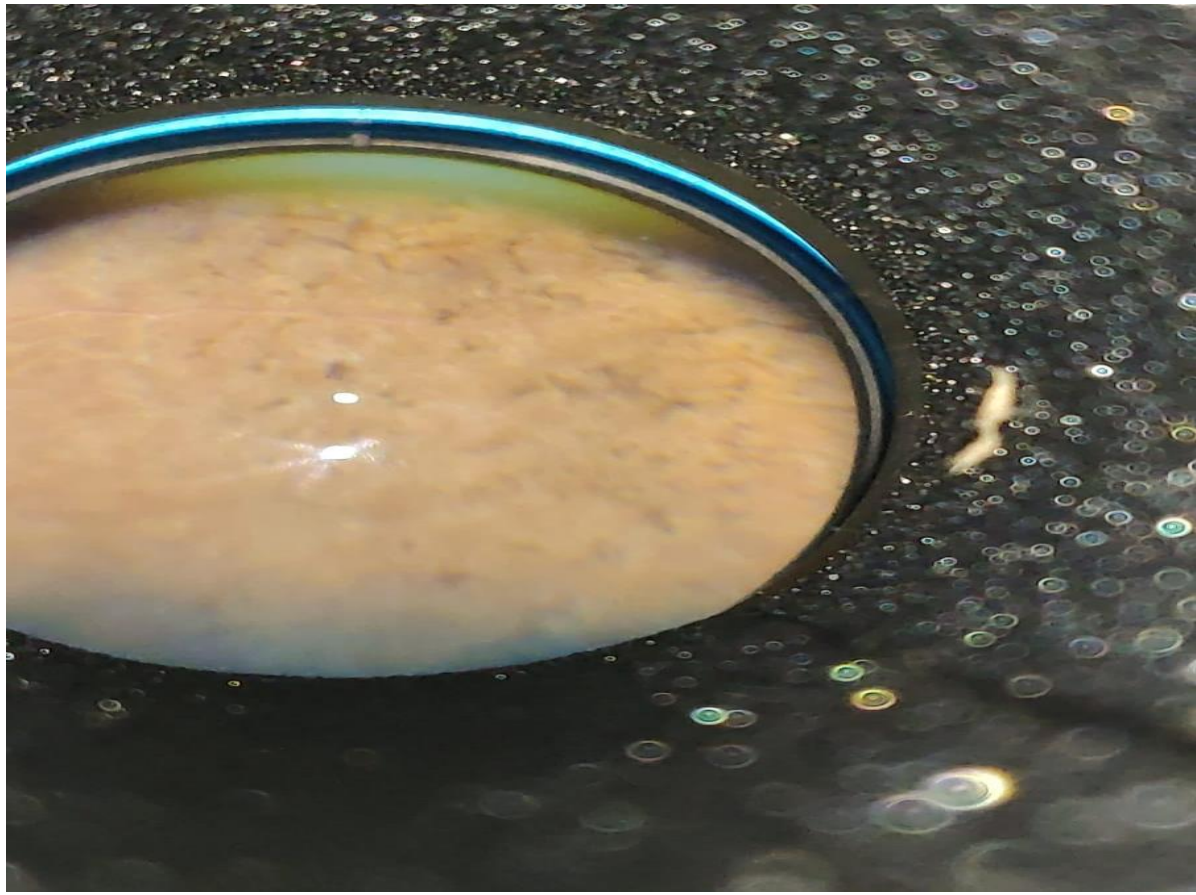
# Working Diagnosis:

- Anaemia of chronic disease with night blindness with bilateral sensory neural hearing loss with ? Acute on chronic kidney disease, likely due to genetic cause ?Alport’s syndrome

- USG A/P:
  - Right kidney (85 x 28mm). CMD maintained.
  - Left kidney (87 x 30 mm). CMD maintained.
  - Bilateral raised cortical echogenicity.
  - Rest, WNL.

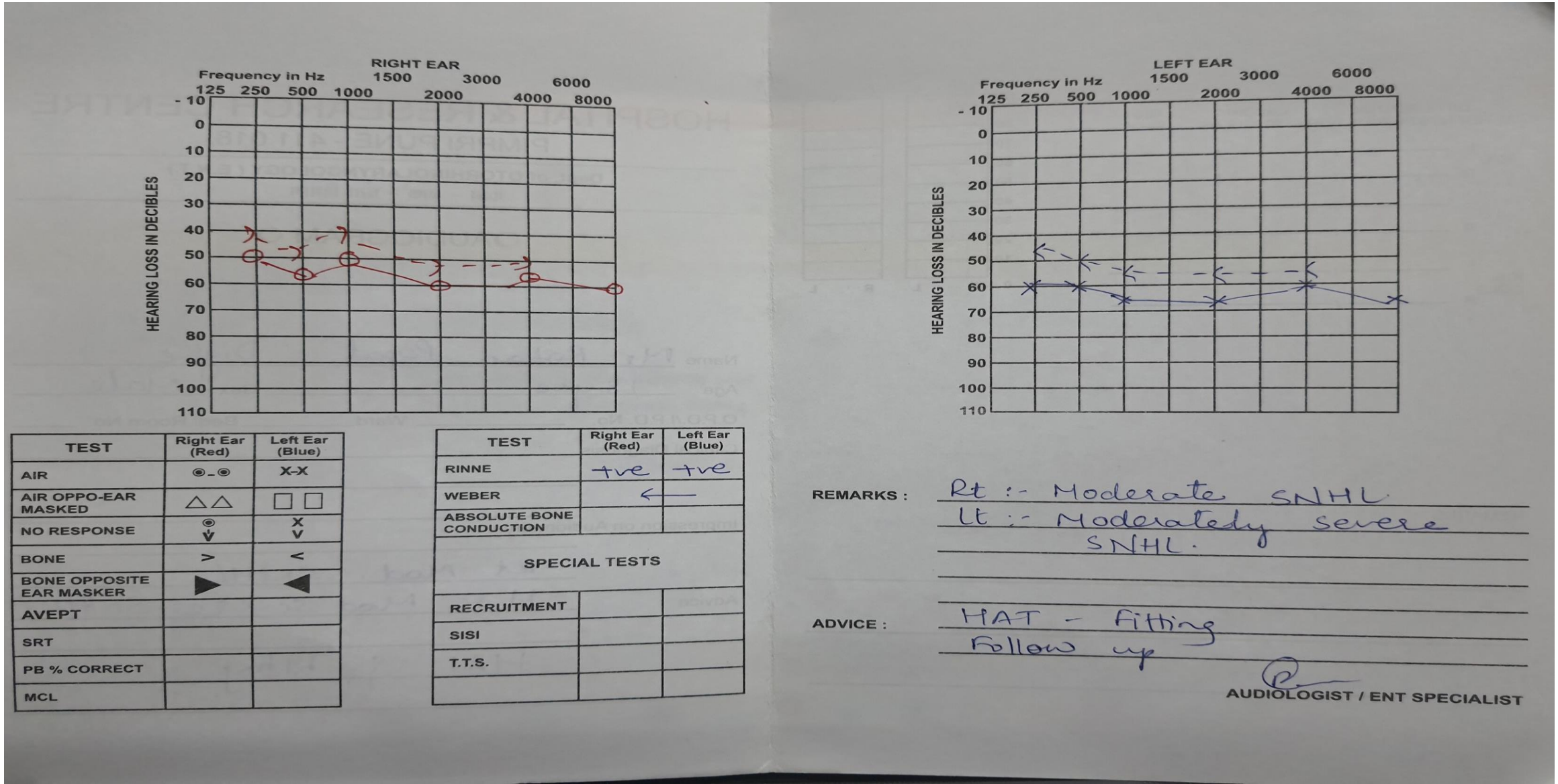
- Nephrology reference:
  - Impression: ? Alports syndrome
  - Advice: Hemodialysis.





- Ophthalmology reference:
  - Both eyes retinitis pigmentosa present.
  - Vitreo-retinal surgery opinion: no active management.

- Pure tone audiometry:
  - Bilateral sensory neural hearing loss.



# TREATMENT:

- Tab Albendazole 400mg stat
- Inj Iron sucrose
- Inj calcium gluconate
- Inj soda bicarbonate as per VBG
- Tab calcium acetate
- Patient was started on Hemodialysis. 3 PCV were transfused during successive HD. Hemodialysis was carried out daily for first 5 days followed by every alternate day for 2 weeks. Patient requires maintenance hemodialysis and is now awaiting permanent catheter placement.

# ALPORT'S SYNDROME:

- Due to defect in type 4 collagen.

- Four forms of the AS are now recognized:
- (1) classic AS, which is inherited as an X-linked disorder with hematuria, sensorineural deafness, and conical deformation of the anterior surface of the lens (lenticonus)
- (2) an X-linked form associated with diffuse leiomyomatosis
- (3) an autosomal recessive form
- (4) an autosomal dominant form.

- X- linked Alport’s:
  - Disease seen only in males. Females are carriers.
  - Involvement of alpha-5 chain on type 4 collagen.
  - Presents with haematuria, sensory neural hearing loss and anterior lenticonus.