

- **Vedantaa institute of Medical Sciences**

- **Department of Paediatrics**

- Dr Bhadbhade
- Assistant professor Dr Mayur Pagare

14 YEARS OLD ,FEMALE PATIENT

ADMITTED ON 26-09-18

WITH CHIEF C/O

MENORRHOEGIA SINCE 2 YEARS.

GENERALISED WEAKNESS.

MENARCHE 2 YEARS AGO,PERIODS IRREGULAR, LASTING FOR OVER 7
DAYS,AND WITH EXCESSIVE BLEEDING PV.

NO H/O ANY BONE PAINS/JOINT PAINS/SWELLING.

NO H/O FEVER.

NO H/O BLEEDING DIATHESIS.

NO H/O ANY PREVIOUS TREATMENT ANYWHERE ELSE IN THE PAST.

NO H/O BLOOD TRANSFUSIONS IN THE PAST.

THIS IS THE FOURTH CHILD ,WITH NO H/O CONSANGUINITY.

OF THE OTHER THREE SIBLINGS ONE MALE,& TWO FEMALES ARE HEALTHY.

O/E,

WT-30KG

HT-150CMS.

VITALS –NORMAL.

NO LYMPHADENOPATHY.

NO BONY TENDERNESS,

PALLOR-+++

NAILS NORMAL

ICTERUS +

NO OEDEMA/ s/o CCF.

NO DENTAL PIGMENTATION.

RS-NORMAL.

CVS-ONLY HEMIC MURMURS +

P.A.-SOFT,NO TENDERNESS,LIVER -1CM,SOFT,NONTENDER.

SPLEEN-2.5 CM.FIRM,NONTENDER.

CNS –LMN 7th N PALSY WHICH THE GRANDMA SAID WAS SINCE BIRTH.

NO OTHER NEUROLOGICAL ABNORMALITY WAS DETECTED.

CLINICALLY THOUGHT TO BE:

?HEMOLYTIC ANEMIA –MOSTLY SICKLE CELL ANEMIA,WITH A COMPONENT OF ASSOCIATED IRON DEFICIENCY IN VIEW OF MENORRHOEGIA OF LONG STANDING.

SO BASELINE INVESTIGATIONS WERE DONE THE REPORTS WERE AS FOLLOWS:

HAEMATOLOGICAL INVESTIGATIONS

No.	Test	Method of testing	Result	Reference range and Units
1.	Haemoglobin	Cell counter	4.1	M: 14 - 17 Gm/dl F: 12 - 16 Gm/dl
2.	Total WBC count		6400	4000 - 11000/ cu mm
3.	Differential count	By smear examination
	Neutrophils		72	40 - 80%
	Lymphocytes		25	20 - 40%
	Eosinophils		01	1 - 6%
	Monocytes		02	2 - 10%
	Basophils		00	0 - 1%
	Immature cells			
4.	Total RBC count	Cell counter	3.2	M: 4.5 - 5.5, F: 3.8 - 4.8 million/ cu mm
5.	RBC Indices	
	Haematocrit (P.C.V.)		15	M: 40 - 50% f: 36 - 40%
	M.C.V.		47	82 - 92 Fl
	M.C.H.		12	27 - 32 Pg
	M.C.H.C.		26	32 - 35%
	R.D.W.		26.9	11.6 - 14%
6.	PLATELET COUNT		1,60,000	1.5 - 4.5 lakhs/ cu mm
7.	Parasites	MP By kit → Negative		
8.	Peripheral smear examination			
9.	E.S.R.	Westergren	114	M: < 10 mm/hr. F: < 15 mm/hr.
10.	Sickling test	Screening test by DTT method		
11.	Blood group & Rh Factor	A +ve (positive)		
12.	BT & CT			
13.	Any other tests			
14.				

HB 4.1GMS%
TLC 6400/cmm
DLC N-72
 L -25
 E -01
 M -02

RBC 3.2 Million/cmm.
PCV 15%
MCV 47
MCH 12
MCHC 26
RDW 26.9
PLATELETS 160000/cmm
MP NEGATIVE.
ESR 114mm at 1hr.

BIOCHEMISTRY INVESTIGATIONS

No.	Test	Result	Reference range and Units
1.	BLOOD SUGAR RBS	139	70-140 mg/dl
	FASTING (FBS)		<125 mg/dl
	FASTING URINE SUGAR (FUS)		
	PPBS		<140 mg/dl
	POST PRANDIAL URINE SUGAR (PPUS)		
2.	BILIRUBIN TOTAL		0.3-1.2 mg /dl
	DIRECT		0.0-0.20 mg /dl
	INDIRECT		0.0-0.70 mg /dl
3.	SGPT (ALT)		<45 IU/L
4.	SGOT (AST)		<38 IU/L
5.	ALP		53-128 IU/L
6.	TOTAL PROTEINS		6.6-8.3 gm /dl
7.	ALBUMIN		3.5-5.2 gm /dl
8.	GLOBULINS		2.3-8.5 gm /dl
	A:G RATIO		
9.	BLOOD UREA		10-45 mg /dl
10.	BUN		8-21 mg /dl
11.	CREATININE	0.9	0.7-1.3 mg /dl
12.	URIC ACID		3.0-7.0 mg/dl
13.	SODIUM		136-145 mEQ /
14.	POTASSIUM		L 3.5-5.1 mEQ /L
15.	CHLORIDE		98-107 mEQ /L
16.	CHOLESTEROL		<200 mg /dl
17.	TRIGLYCREIDE		<170 mg /dl >
18.	HDL		50 mg /dl
19.	LDL		<130 mg /dl
20.	VLDL		0.0 - 35
21.	CHO:HDL		0.0 - 5.0
22.	CALCIUM		8.5-11 mg /dl
23.	AMYLASE		0-80 U/L
24.	LIPASE		13-60 U/L
25.			

RBSL 139mg%

SE.CREAT. 0.9mg%

HAEMATOLOGICAL INVESTIGATIONS

No.	Test	Method of testing	Result	Reference range and Units
1.	Haemoglobin	Cell counter	5.1	M: 14 - 17 Gm/dl F: 12 - 16 Gm/dl
2.	Total WBC count		8,200	4000 - 11000/cu mm
3.	Differential count	By smear examination
	Neutrophils		85	40 - 80%
	Lymphocytes		10	20 - 40%
	Eosinophils		03	1 - 6%
	Monocytes		02	2 - 10%
	Basophils		00	0 - 1%
	immature cells		-	
4.	Total RBC count	Cell counter	3.8	M: 4.5 - 5.5, F: 3.8 - 4.8 million/cu mm
5.	RBC indices	
	Haematocrit (P.C.V.)		19	M: 40 - 50% f: 36 - 40%
	M.C.V.		50	82 - 92 Fl
	M.C.H.		13	27 - 32 Pg
	M.C.H.C.		26	32 - 35%
	R.D.W.		27.9	11.6 - 14%
6.	PLATELET COUNT		8,61,000	1.5 - 4.5 lakhs/cu mm
7.	Parasites			
8.	Peripheral smear examination		RBC - Microcytic Hypochromic Anisopoikilocytosis teardrop cells, elliptocytes. WBC - WNL. Plt - Increased on PS Parasite -	
9.	E.S.R.	Westergren		M: < 10 mm/hr. Negativity F: < 15 mm/hr. Con 12
10.	Sickling test	Screening test by DTT method		
11.	Blood group & Rh Factor			
12.	BT & CT			
13.	Any other tests			
14.				

HB 5.1GMS%
TLC 8200/cmm
DLC N-85

L -10

E -03

M -02

RBC 3.8 Million/cmm.

PCV 19%

MCV 50

MCH 13

MCHC 26

RDW 27.9

PLATELETS 861000/cmm

MP NEGATIVE.

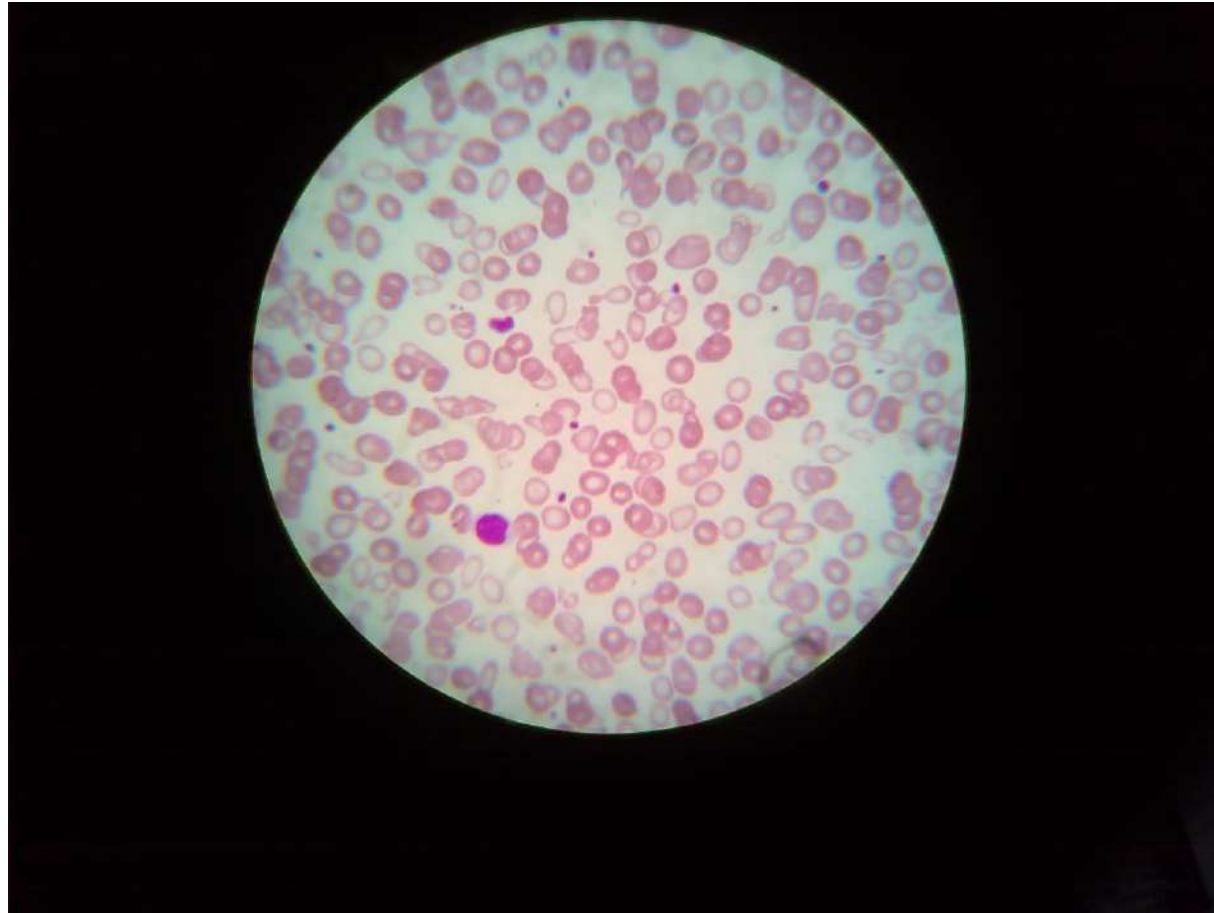
ESR 114mm at 1hr.

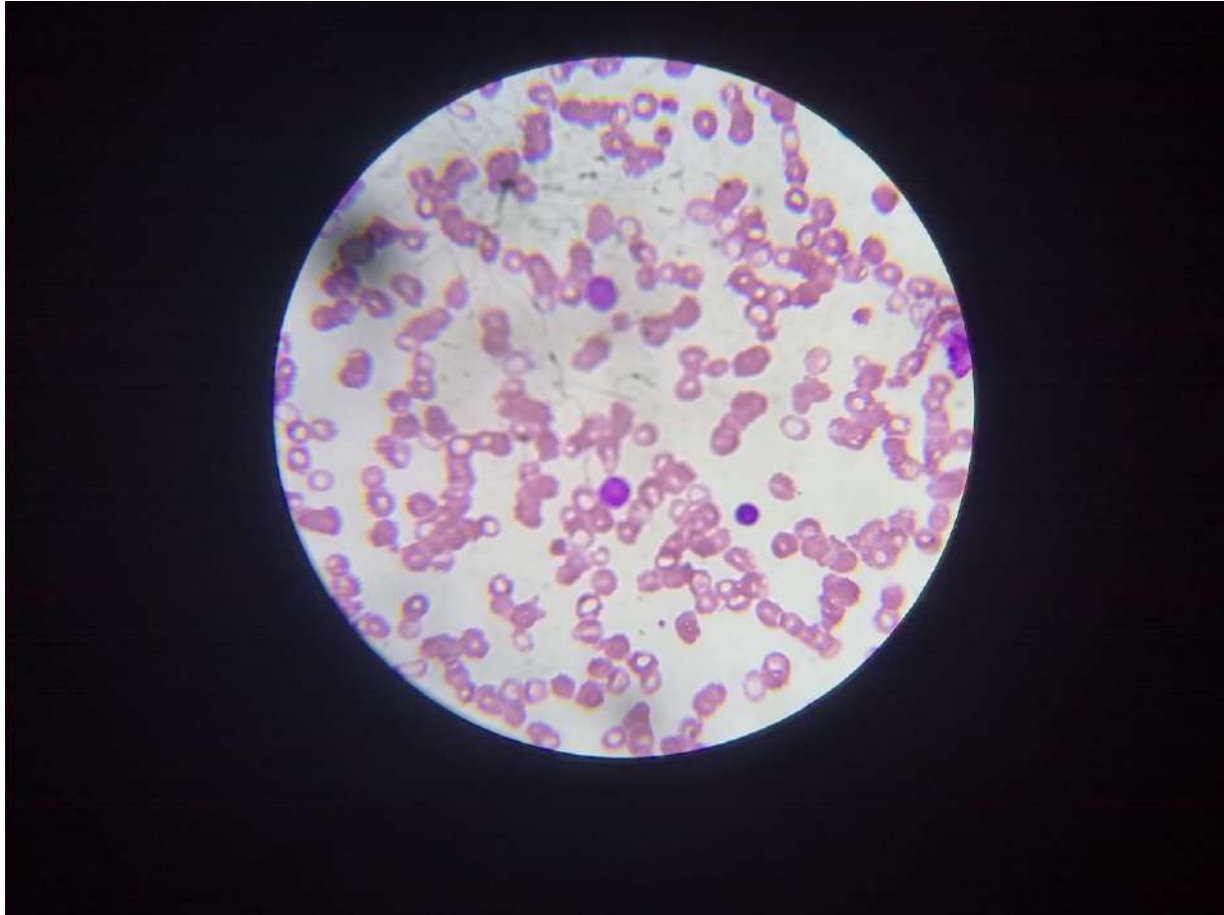
RBCs Microcytic, Hypochromic,
Anisocytosis+Poikilocytosis+
Teardrop cells+, elliptocytosis+No
Target Cells/Spherocytes.

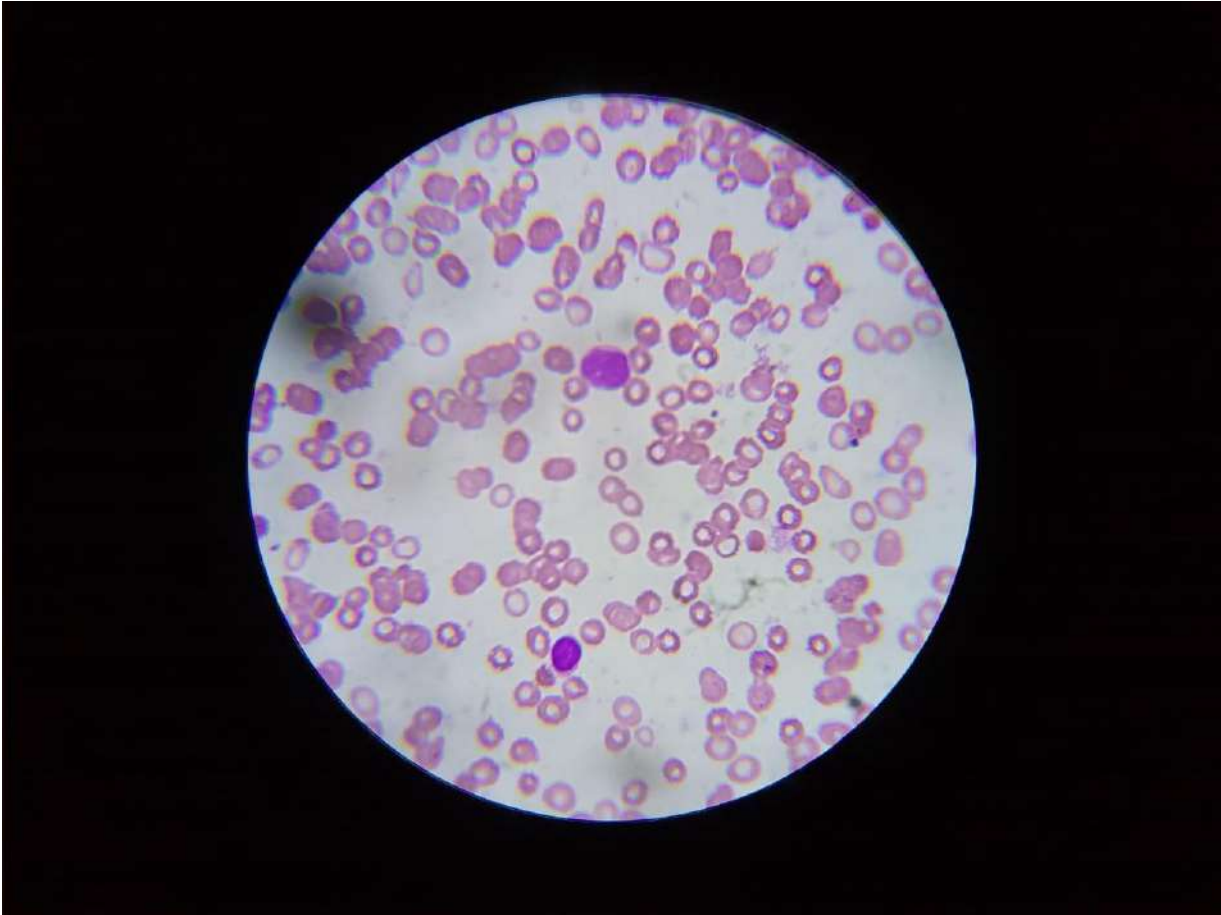
WBCs-WNL

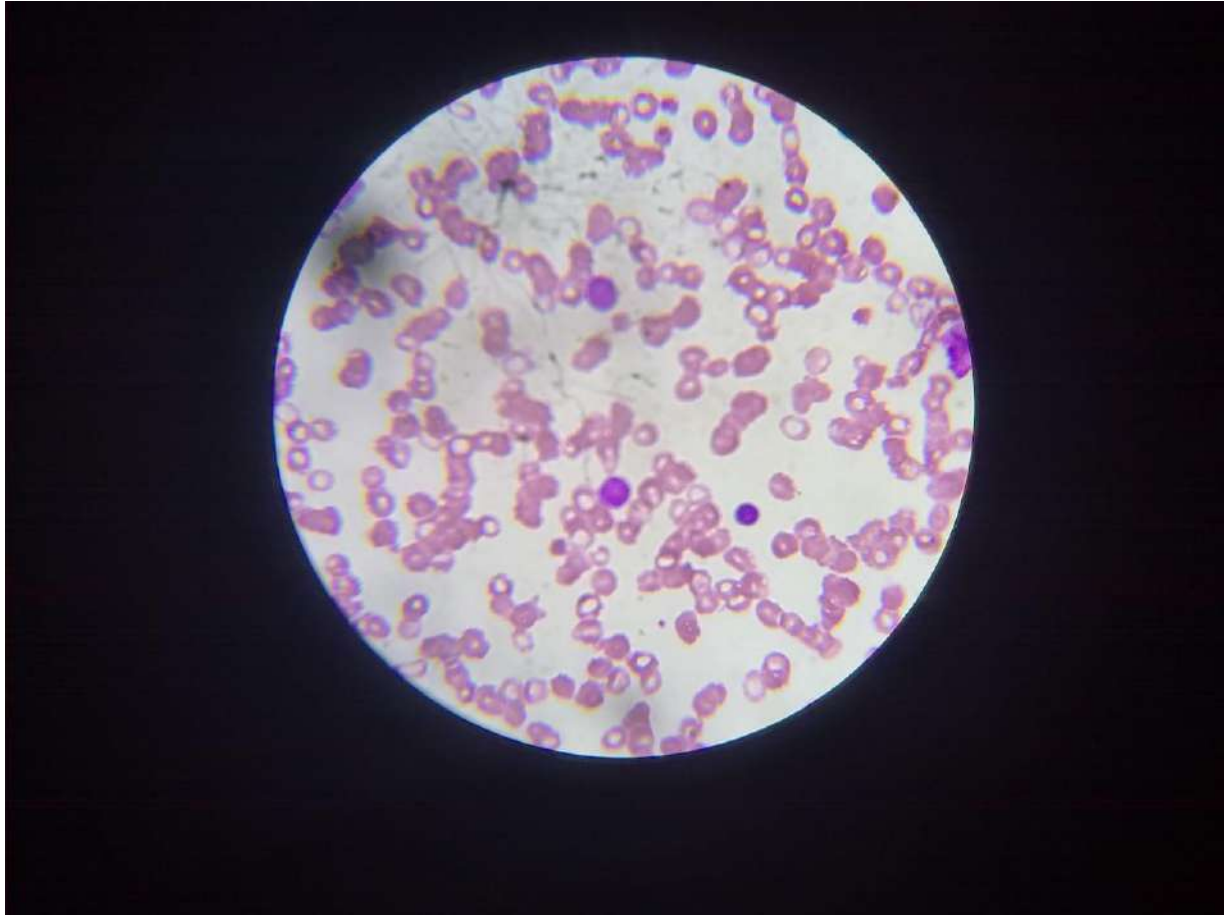
Platelets-Increased.

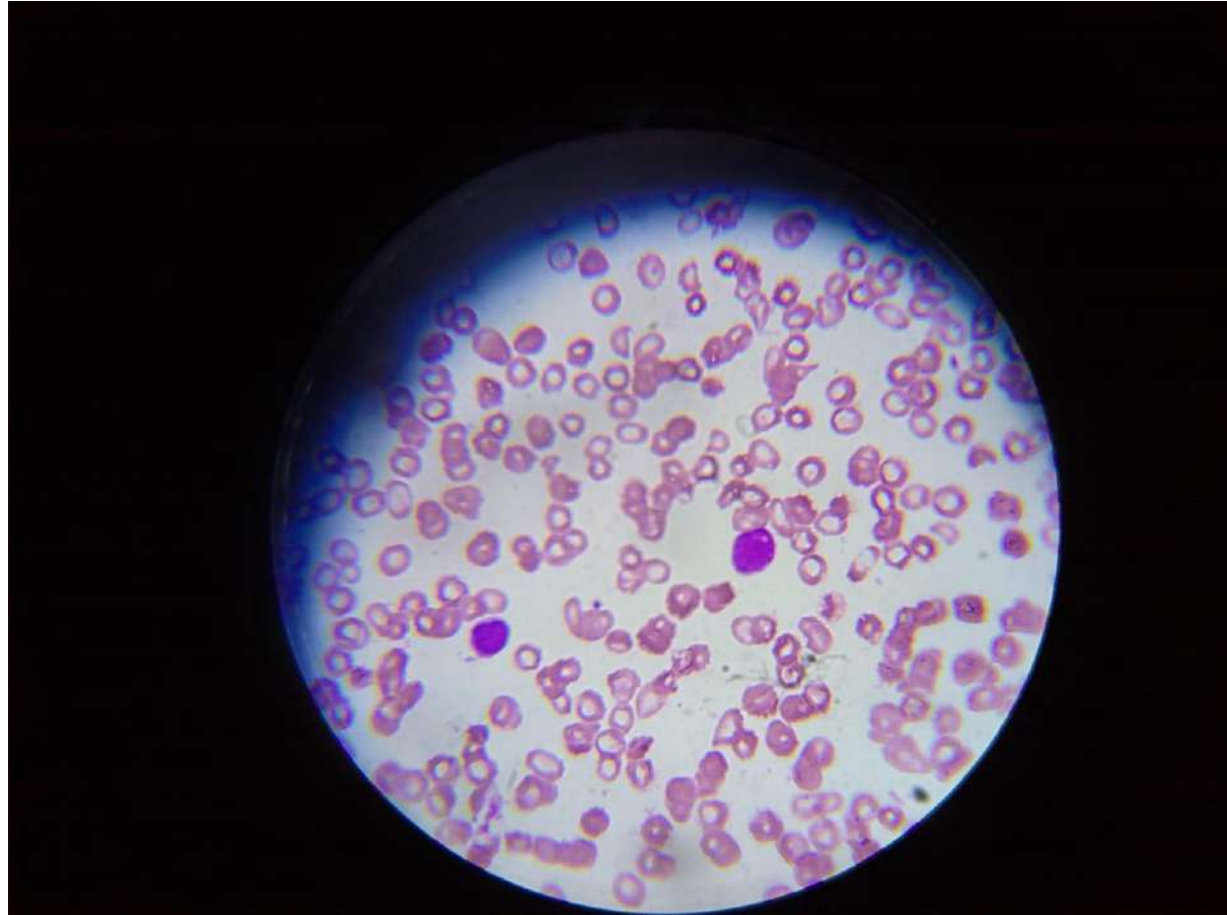
Peripheral Smear

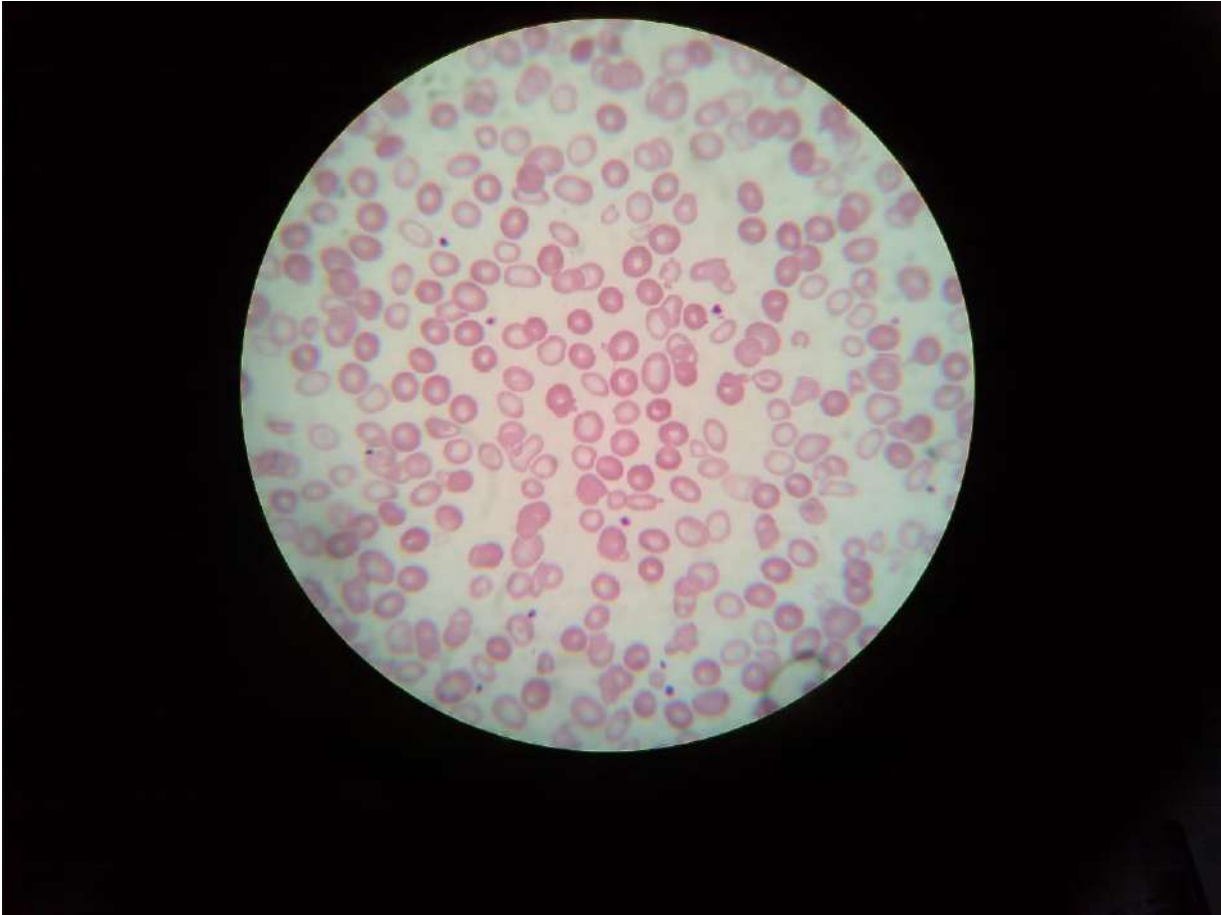












CBC POST TRANSFUSION

HAEMATOLOGICAL INVESTIGATIONS			
No.	Test	Method of testing	Result
1.	Haemoglobin	Cell counter	10.1
2.	Total WBC count	By smear examination	6,800
3.	Differential count	By smear examination	
	Neutrophils		58
	Lymphocytes		38
	Eosinophils		02
	Monocytes		02
	Basophils		00
	Immature cells		00
4.	Total RBC count	Cell counter	5.5
5.	RBC Indices		
	Haematocrit (P.C.V.)		34
	M.C.V.		61
	M.C.H.		18
	M.C.H.C.		29
	R.D.W.		37.1
6.	PLATELET COUNT		3,07,000
7.	Parasites		
8.	Peripheral smear examination		Hypochromasia +, microcytosis +, Anisopoikilocytosis +, pencil cells +, Platelets adequate on smear, macrocytes +
	E.S.R.	Westergren	114 mm/hr.
	Sickling test	Screening test by DTT method	No sickling
Blood group & Rh Factor			
BT & CT			
Any other tests			
Retic: 2% Dimorphic Anemia.			

HB 10.1GMS%
TLC 6800/cmm
DLC N-58
 L -38
 E -02
 M -02

RBC 5.5 Million/cmm.
PCV 34%
MCV 61
MCH 18
MCHC 29
RDW 37.1
PLATELETS 307000/cmm
MP NEGATIVE.
ESR 114mm at 1hr.
 Hypochromia, microcytosis, anisopoikilocytosis, macrocytosis, RETIC-2%

URINE ANALYSIS

PHYSICAL EXAMINATION	Quantity	10 ml
	Colour	Yellow
	Appearance	Hazy
	Reaction	Alkaline
	Specific gravity	1.010
CHEMICAL EXAMINATION	Albumin	Present (Trace)
	Sugar	Absent
	Ketone bodies	Absent
	Bile salts & Bile pigments	Absent
	Urobilinogen	Absent
	Occult Blood	Absent
	MICROSCOPIC EXAMINATION	Pus cells
RBCs		Absent
Epithelial cells		4-5/hpf
Casts		Absent
Crystals		Absent
Amorphous deposits		Absent
Bacteria		Absent
OTHER FINDINGS		
PREGNANCY TEST	Strip test for detection of HCG in urine	

URINE ANALYSIS

Color	Yellow
Albumin	Trace
Sugar	Absent
Bile Salts	Absent
Bile Pigm.	Absent
Urobilino.	Absent
OBT	Negative
Pus Cells	6-7/hpf
RBCs	Absent
Epi.Cells	4-5/hpf
Casts	Absent
Crystals	Absent

HAEMATOLOGICAL INVESTIGATIONS

No.	Test	Method of testing	Result	Reference range and Units
1.	Haemoglobin	Cell counter		M: 14 - 17 Gm/dl F: 12 - 16 Gm/dl
2.	Total WBC count			4000 - 11000/cu mm
3.	Differential count	By smear examination
	Neutrophils			40 - 80%
	Lymphocytes			20 - 40%
	Eosinophils			1 - 6%
	Monocytes			2 - 10%
	Basophils			0 - 1%
	immature cells			
4.	Total RBC count	Cell counter		M: 4.5 - 5.5, F: 3.8 - 4.8 million/cu mm
5.	RBC indices			...
	Haematocrit (P.C.V.)			M: 40 - 50% F: 36 - 40%
	M.C.V.			82 - 92 Fl
	M.C.H.			27 - 32 Pg
	M.C.H.C.			32 - 35%
	R.D.W.			11.6 - 14%
6.	PLATELET COUNT			1.5 - 4.5 lakhs/cu mm
7.	Parasites			
8.	Peripheral smear examination			
9.	E.S.R.	Westergren		M: < 10 mm/hr. F: < 15 mm/hr.
10.	Sickling test	Screening test by DTT method - <i>Negative</i>		
11.	Blood group & Rh Factor			
12.	BT & CT			
13.	Any other tests			
14.				

SICKLING TEST- SCREENING TEST –
NEGATIVE
RETIC COUNT -1%

BIOCHEMISTRY INVESTIGATIONS

No.	Test	Result	Reference range and Units
1.	BLOOD SUGAR RBS		70-140 mg/dl
	FASTING (FBS)		<126 mg/dl
	FASTING URINE SUGAR (FUS)		
	PPBS		<140 mg/dl
	POST PRANDIAL URINE SUGAR (PPUS)		
2.	BILIRUBIN TOTAL	4.2	0.3-1.2 mg/dl
	DIRECT	0.3	0.0-0.20 mg/dl
	INDIRECT	3.9	0.0-0.70 mg/dl
3.	SGPT (ALT)	10	<45 IU/L
4.	SGOT (AST)	22	<38 IU/L
5.	ALP		53-128 IU/L
6.	TOTAL PROTEINS		6.5-8.3 gm/dl
7.	ALBUMIN		3.5-5.2 gm/dl
8.	GLOBULINS		2.3-3.5 gm/dl
	A:G RATIO		
9.	BLOOD UREA		10-45 mg/dl
10.	BUN		8-21 mg/dl
11.	CREATININE		0.7-1.3 mg/dl
12.	URIC ACID		3.0-7.0 mg/dl
13.	SODIUM		136-145 mEq/L
14.	POTASSIUM		3.5-5.1 mEq/L
15.	CHLORIDE		98-107 mEq/L
16.	CHOLESTEROL		<200 mg/dl
17.	TRIGLYCEIDE		<170 mg/dl>
18.	HDL		50 mg/dl
19.	LDL		<130 mg/dl
20.	VLDL		0.0 - 35
21.	CHO:HDL		0.0 - 5.0
22.	CALCIUM		8.5-11 mg/dl
23.	AMYLASE		0-80 U/L
24.	LIPASE		13-60 U/L
25.			

BILIRUBIN TOTAL - 4.2mgm%
DIRECT - 0.3mgm%
INDIRECT- 3.9mgm%
SGPT - 10 IU/L
SGOT - 22 IU/L

ULTRASOUND ABDOMEN & PELVIS

LIVER: 12.4 cm (N) Echo texture.

GALL BLADDER: well distended (N).

SPLEEN: 8.5 cm

PANCREAS: (N)

RT. KIDNEY: 9.1 x 3.4 cm (N) CMD

LT. KIDNEY: 9.7 x 3.8 cm (N) CMD

U. BLADDER: well distended (N)

UTERUS/PROSTATE: - RTO - 3.2 x 1.7 cm
Uterus - 6.4 x 3.0 x 4 cm
LTO - 4.0 x 2.0 cm

IMPRESSION: ET - 7 mm

Normal Scan


Radiology Resident

USG ABDOMEN- NORMAL SCAN

HAEMATOLOGICAL INVESTIGATIONS

No.	Test	Method of testing	Result	Reference range and Units
1.	Haemoglobin	Cell counter	9.4	M: 14 - 17 Gm/dl F: 12 - 16 Gm/dl
2.	Total WBC count		9,300	4000 - 11000/ cu mm
3.	Differential count	By smear examination		
	Neutrophils		67	40 - 80%
	Lymphocytes		24	20 - 40%
	Eosinophils		03	1 - 6%
	Monocytes		05	2 - 10%
	Basophils		01	0 - 1%
	Immature cells		-	
4.	Total RBC count	Cell counter	5.7	M: 4.5 - 5.5, F: 3.8 - 4.8 million/ cu mm
5.	RBC Indices			
	Haematocrit (P.C.V.)		35	M: 40 - 50% f: 36 - 40%
	M.C.V.		61	82 - 92 fl
	M.C.H.		18	27 - 32 Pg
	M.C.H.C.		29	32 - 35%
	R.D.W.		27	11.6 - 14%
6.	PLATELET COUNT		Plt adequate on smear 15-16 Lakh/cmm	
7.	Parasites		Large platelet present. manual in 3.5 Lakh	
8.	Peripheral smear examination		Hypo + micro + Anisoblast + macro + Teardrop cells. few	
9.	E.S.R.	Westergren		M: < 10 mm/hr. F: < 15 mm/hr.
10.	Sickling test	Screening test by DTT method		
11.	Blood group & Rh Factor			
12.	BT & CT			
13.	Any other tests			
14.				

POST TRANSFUSION CBC

HB	9.4GMS%
TLC	9300/cmm
DLC	N-67 L -24 E -03 M -05 B - 01
RBC	5.7 Million/cmm.
PCV	35%
MCV	61
MCH	18
MCHC	29
RDW	37
PLATELETS	350000/cmm

RBCs - HYPO+, MICRO+,MACRO+,TEARDROP CELLS FEW, LARGE PLATELETS on PS.

BIOCHEMISTRY INVESTIGATIONS

No.	Test	Result	Reference range and Units
1.	BLOOD SUGAR		
	RBS		70-140 mg/dl
	FASTING (FBS)		<126 mg/dl
	FASTING URINE SUGAR (FUS)		
	PPBS		<140 mg/dl
	POST PRANDIAL URINE SUGAR (PPUS)		
2.	BILIRUBIN TOTAL	2.0	0.3-1.2 mg/dl
	DIRECT	0.7	0.0-0.20 mg/dl
	INDIRECT	1.3	0.0-0.70 mg/dl
3.	SGPT (ALT)	27	<45 IU/L
4.	SGOT (AST)	41	<38 IU/L
5.	ALP	72	53-128 IU/L
6.	TOTAL PROTEINS	7.5	6.5-8.3 gm/dl
7.	ALBUMIN	4.0	3.5-5.2 gm/dl
8.	GLOBULINS		2.3-8.5 gm/dl
	A.G RATIO		
9.	BLOOD UREA		10-45 mg/dl
10.	BUN		8-21 mg/dl
11.	CREATININE		0.7-1.3 mg/dl
12.	URIC ACID		3.0-7.0 mg/dl
13.	SODIUM		135-145 mEq/L
14.	POTASSIUM		3.5-5.1 mEq/L
15.	CHLORIDE		98-107 mEq/L
16.	CHOLESTEROL		<200 mg/dl
17.	TRIGLYCERIDE		<170 mg/dl >
18.	HDL		50 mg/dl
19.	LDL		<130 mg/dl
20.	VLDL		0.0 - 35
21.	CHO:HDL		0.0 - 5.0
22.	CALCIUM		8.5-11 mg/dl
23.	AMYLASE		0-80 U/L
24.	LIPASE		13-60 U/L
25.			

ON 05-10-18

BILIRUBIN TOTAL 2.00 mgm%
DIRECT 0.70 mgm%
INDIRECT 1.30 mgm%
SGPT 27 IU/L
SGOT 41 IU/L
ALP 72 IU/L
TOTAL PROTEINS 7.5 Gms%
ALBUMIN 4.00 Gms%



(EDTA Whole Blood)

<u>Investigation</u>	<u>Observed Value</u>	<u>Unit</u>	<u>Biological Reference Interval</u>
Foetal Haemoglobin (HbF)	0.3	%	0.0-2.0
Haemoglobin A0 (Hb A0)	97.7	%	94.3-98.5
Haemoglobin A2 (HbA2)	2.0	%	1.5-3.7
I	Kindly provide History of blood transfusion for a definite opinion.		
<u>Method</u>	HPLC		
I			

FROM METROPOLIS LABS

04-10-18

FOETAL HB (HBF) 0.3%
HBA0 (HBAo) 97.7%
HBA2 02%

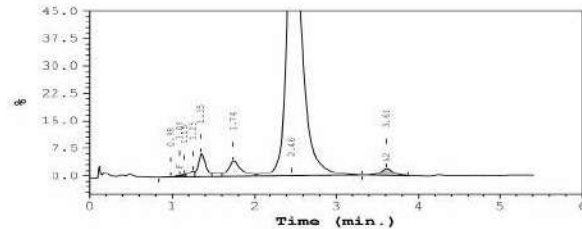
<u>Patient Data</u>	<u>Analysis Data</u>
Sample ID: 119521647	Analysis Performed: 01/10/2018 17:22:52
Patient ID: 11191174321	Injection Number: 21100
Name: MUKTI J M	Run Number: 122
Physician:	Rack ID:
Sex:	Tube Number: 3
DOB:	Report Generated: 02/10/2018 14:46:13
Comments:	Operator ID:

Peak Name	Calibrated Area %	Area %	Retention Time (min)	Peak Area
Unknown	---	0.0	0.98	765
F	0.3	---	1.09	4508
Unknown	---	0.3	1.15	4289
Unknown	---	0.8	1.25	12904
P2	---	3.6	1.35	59705
P3	---	4.3	1.74	71229
Ao	---	98.7	2.46	1474010
A2	2.0	---	3.61	33818

Total Area: 1,661,229

F Concentration = 0.3 %
A2 Concentration = 2.0 %

Analysis comments:



Investigation	Observed Value	Unit	Biological Reference Interval
Iron Studies, Serum (Serum)			
I (Ferene)	347	µg/dL	2
T (Ferene)		µg/dL	2
U (Calculated)	56.00	µg/dL	1
T (Calculated)	86	%	1
Interpretation			

FROM METROPOLIS LABS
04-10-18

SE.IRON- 347 Microgm/DL. REF-25-102

TIBC - 403 Microgm./DL REF-250-420

UIBC - 56 Microgm/DL REF-120-470

TRAN.SAT. 86% REF-14-50%

T	I	A	I	H
	anaemia	Chronic disease		(Especially Trait)
Serum Iron	Decreased	Decreased	Increased	Normal
S Binding Capacity	I	D Normal	I	N
% Saturation	D	D Normal	I	N
S	I	D Normal	D	N
Serum Ferritin	Decreased	Increased	Increased or Normal	Normal
S Transferrin receptor	I	N	D	N
Serum Hcpcidin	Normal	Increased	Normal	Normal

A
1
2

- Brief Summary of the case:

A 14 yrs old girl with menorrhagia of 2 years duration, not having taken any treatment before was detected in a diagnostic camp with severe anemia, mild jaundice, and a congenital LMN type 7th N palsy, and was admitted for investigations.

Clinically thought to be a c/o ? severe anemia due to Hemolytic anemia(?Sickle Cell- in view of the community she came from), acholuric jaundice (because of normal LFTs). Probably with an element of associated iron deficiency too! (in view of her longstanding untreated menorrhoea).

Routine baseline studies supported this suspicion as CBC showed a **severely microcytic hypochromic anemia with marked increase in RDW but with severe anisopoikilocytosis.**

Though screening test for sickling was negative, and retic count was normal, in view of her jaundice, sample was sent for Hb Electrophoresis.

Iron studies were also sent in view of severe microcytic, hypochromic anemia, and increased RDW.

By the time she was given Blood Transfusion after sending samples for these studies, her jaundice had almost subsided. LFTs continued to be normal.

- Her Hb Electrophoresis report came absolutely normal.
- The twist in the tale to our surprise was however, her iron studies report which showed
 - 1) **Markedly increased Serum Iron levels- SE.IRON- 347 MicroGms/DL.**
With a Ref range of -25-102micrograms/DL.
 - 2) **TIBC - 403 MicroGms./DL**
With a Ref range -250-420MicroGms/DL
 - 3) **UIBC - 56MicoGms./DL With a Ref range-120-470MicroGms/DL.**
 - 4) **TRAN.SATU.86% Ref range -14-50%**

CAUSES of MICROCYTIC HYPOCHROMIC ANEMIA

- 1) IRON DEFICIENCY ANEMIA
- 2) BETA THALASSEMIA MAJOR
- 3) THALASSEMIA MINOR.
- 4) OTHER HAEMOGLOBINOPATHIES. –HB E,HB H Etc.
- 5) ANEMIA OF CHRONIC DISEASES
- 6) LEAD POISONING
- 7) SIDEROBLASTIC ANEMIA.
- 8) CONGENITAL ABSENCE OF IRON BINDING PROTEIN (ATRANSFERINEMIA).

.SIDEROBLASTIC ANEMIA.

*A RARE MICROCYTIC HYPOCHROMIC ANEMIA WHICH ALSO HAS MIXED NORMAL RBCs GIVING A PICTURE OF DIMORPHIC ANEMIA. CHARACTERISED BY EXTREMELY HIGH RDWs.

*SERUM IRON LEVELS ARE INCREASED,

TRANSFERIN SATURATION IS INCREASED GIVING LOW UIBCs.

TIBC MAY BE NORMAL OR HIGH NORMAL.

- RESULTS FROM IMPAIRED HEME SYNTHESIS LEADING TO RETENTION OF IRON IN MITOCHONDRIA. SEEN IN THE MARROW AS NUCLEATED RBCs WITH PERINUCLEAR IRON GRANULES THAT RESULT IN TO RING SIDEROBLASTS IN CONTRAST TO DIFFUSE CYTOPLASMIC FERRITIN GRANULES IN NORMAL SIDEROBLASTS IN THE BONE MARROW.

*SIDEROBLASTIC ANEMIAS MAY BE CONGENITAL OR ACQUIRED.

CONGENITAL SIDEROBLASTIC ANEMIAS:

X LINKED (still may be seen in females due to skewed lyonization), AUT. DOMINANT, SPORADIC.

RESULTS FROM ABNORMALITIES OF 5 AMINOLEVULINIC ACID SYNTHETASE REQD. FOR HEME SYN.

IMPORTANT COFACTOR FOR THIS REACTION IS PYRIDOXAL PHOSPHATE. MANY DIFF. MUTATIONS.

IF SEVERE, MAY BE SEEN IN INFANCY OR EARLY CHILDHOOD, BUT Milder forms may not become apparent until early adulthood or later.

CLINICALLY PRESENTS AS PALLOR, ICTERUS, SPLENOMEGALY, & MAY BE, HEPATOMEGALY.

SOME OF THESE CASES MAY MANIFEST HEMATOLOGIC RESPONSE TO PHARMACOLOGIC DOSES OF PYRIDOXIN.

IRON OVERLOAD MAY LEAD TO DIABETES MELLITUS AND HEPATIC DYSFUNCTION.

ANEMIA MAY BE MILD OR MAY NEED BLOOD TRANSFUSIONS.

STEM CELL TRANSPLANTATION MAY BE REQD.

PEARSON SYNDROME: CONGENITAL SIDEROBLASTIC ANEMIA WITH NEUTROPENIA, & THROMBOCYTOPENIA.

- **ACQUIRED SIDEROBLASTIC ANEMIA:**
- DUE TO ALCOHOLISM, PYRIDOXIN DEFICIENCY, &
- DRUGS (chloramphenicol, penicillamine, linezolid, tetracyclines, progesterones, chemotherapeutics)

- A WORD ABOUT CONGENITAL LMN TYPE 7th N PALSY.

THIS IS USUALLY A COMPRESSION NEUROPATHY DUE TO FORCEPS APPLICATION, BUT THAT IS TEMPORARY.

IN RARE CIRCUMSTANCES, IT COULD BE DUE TO DEVELOPMENTAL DEFECTS IN THE PONS AND MEDULLA OBLONGATA, OR VASCULAR ABNORMALITIES LIKE CALCIFIED INFARCTS IN TEGMENTUM.

IN MOEBIUS SYNDROME IT IS BILATERAL.

SHOULD BE HOWEVER DIFFERENTIATED FROM CONGENITAL ASYMMETRIC CRYING FACIES DUE TO ABNORMALITIES OF DAOM.