* بسم الله الرحمن الرحيم

9 YEARS OLD FEMALE WITH THROMBOCYTOPENIA

BY

DR. HIBA RAFI AL-ATRACKJEE

SUPERVISED BY

DR. MUNA KASHMOULAH

NAME : Marwa Ausamah Ahmed Hassan. *

SEX : female.

AGE : 9 years old.

ADDRESS: Mosul , Al-farooq st.

CHEIF COMPLAIN: Pallor noticed by her family.

HISTORY OF PRESENT ILLNESS : the condition as a whole discovered accidently when her relative (who working in the lab) noticed pallor associated with flu like illness and mild fever ,doing C.B.C for her and discovered low Hb and severe low platelet count .

On examination : 9 years old female short stature, small for age, conscious ,pale ,afebrile ,looking healthy , active child with normal cognitive function ,no any skin pigmentation ,no red discoloration , no bleeding ,normal hearing, have scar at right hand (of an accessory thumb) , her height 120 cm (below normal growth chart). her weight 23 kg(consider as just normal according to growth chart .

REVIEW OF OTHER SYSTEM: nothing significant

P.M.H. : history of red discoloration before six years distributed allover the body then self limiting (as her family said).

P.S.H. : history of rudimentary thumb of right hand operated on before one years of age.

Drug history : mother take TETRACYCLIN drug in early pregnancy when she was pregnant with Marwa.

Family history : mother had three sons and two daughters all are healthy and no congenital abnormality (except Marwa), her mother and father are relative.

Investigation : **C.B.C.** 29 / 5 / 2007

Hb : 115 g/L		110-140 g/L		
PCV : 0.36 L/L		0.34-0.44 L/L		
MCHC : 31.9 g/dl		31.0-35.0 g/dl		
WBC : 7.5 $\times 10^9$/L		4_11 $\times 10^9$/L		
N → 45%	Absol count :	3.4	(2-7 $\times 10^9$/L)	40 -80%
L → 53 %	:	4	(1-3$\times 10^9$/L)	20 - 40%
M → 2 %	:	0.1	(0.2_1 $\times 10^9$/L)	2 - 10%
E → 0 %	:	0	(0.02 _0.5 $\times 10^9$/L)	1 - 6%
B → 0 %	:	0	(0.02_0.1 $\times 10^9$/L)	0 - 1%
PLATELET : 155 $\times 10^9$/L		200_490 $\times 10^9$/L		

BLOOD CELLS MORPHOLOGY :

RBC: NORMOCHROMIC NORMOCYTIC RBC.

WBC : ALL ARE MATURE.

PLATELET : REDUCED IN FILM.

investigation : C.B.C 5/ 1 / 2013

Hb : 101 g/L	115-155	
PCV : 0.29 L/L	0.35-0.45	
MCV:100 FL	83_101	
MCH:27 Pg	25_33	
MCHC : 34.8 g/dl	31.0-37.0	
Retic : 0.5 %	0.5 - 2.5	
WBC : 4.9 $\times 10^9$/L	5 - 13	
N → 27 %	40 - 75	absolute count:1.32 (2_8 $\times 10^9$/L)
L → 67 %	20 - 45	3.28 (1_5 $\times 10^9$/L)
M → 5 %	2 - 10	0.2(0.2_1 $\times 10^9$/L)
E → 1 %	1 - 6	0.04(0.02_0.5 $\times 10^9$/L)
B → 0 %	0 -1	0.02_0.1 $\times 10^9$/L
PLATELET : 33 $\times 10^9$/L	170 - 450 $\times 10^9$/L	

ESR : 30 mm/hr

RBC: NORMOCHROMIC NORMOCYTIC RBC with macrocytic RBC.

WBC : neutropenia , ALL ARE MATURE.

PLATELET : severely reduced in film.

investigation : C.B.C 6/ 4 / 2013

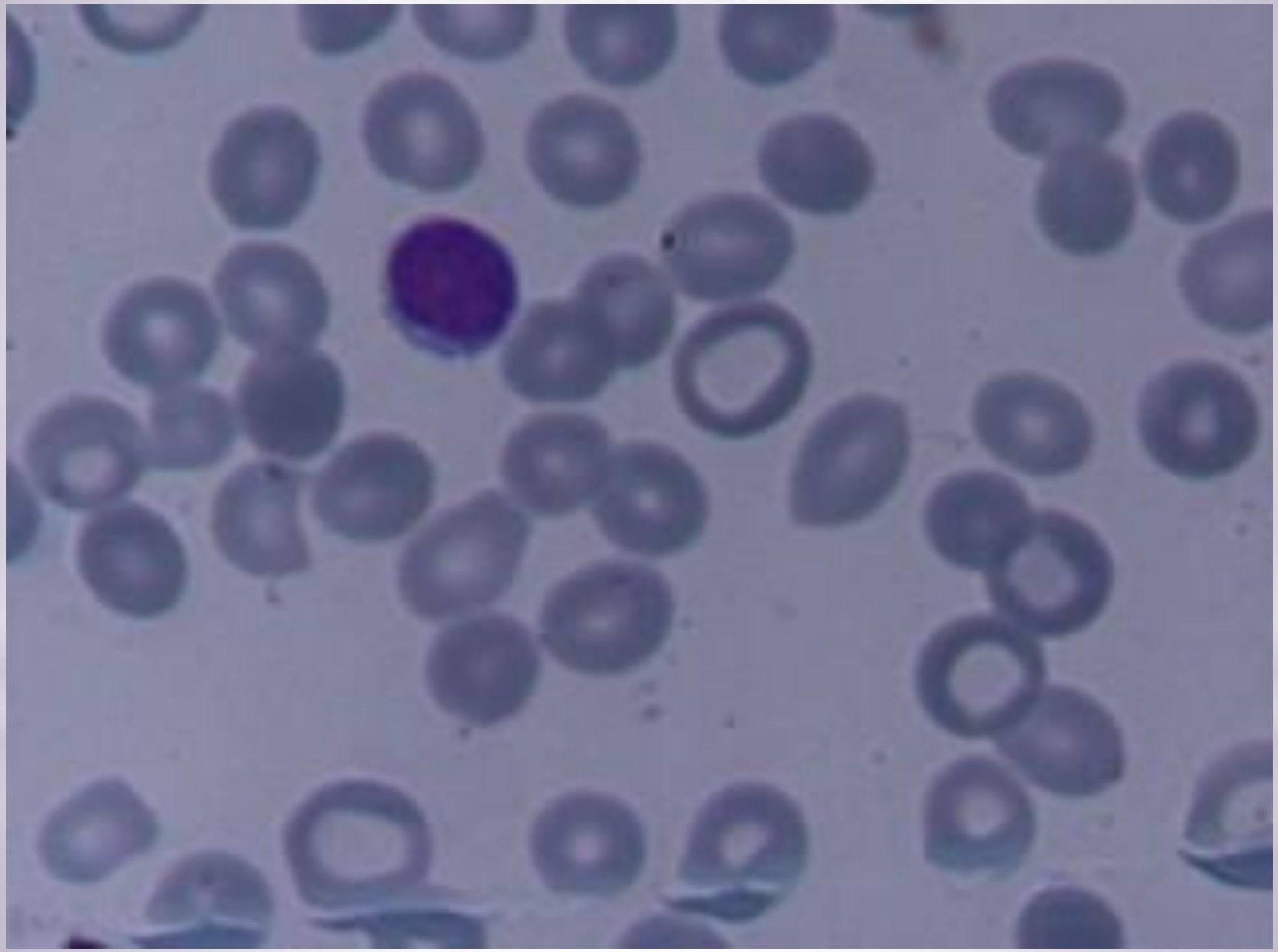
Hb :	8.2 g/L	115-155	
PCV :	0.25 L/L	0.35-0.45	
MCV:	104 FL	83_101	
MCH:	34.2 Pg	25_33	
MCHC:	33.7 g/dl	31.0-37.0	
Retic:	0.5 %	0.5 - 2.5	
WBC:	3.4 $\times 10^9$/L	5 - 13	
N →	17 %	40 - 75	absolute count: 0.58 (2_8 $\times 10^9$/L)
L →	76 %	20 - 45	2.58 (1_5 $\times 10^9$/L)
M →	6 %	2 - 10	0.2(0.2_1 $\times 10^9$/L)
E →	0.2 %	1 - 6	0.007(0.02_0.5 $\times 10^9$/L)
B →	0.8 %	0 -1	0.027 (0.O2_0.1 $\times 10^9$/L)
PLATELET :	19.5 $\times 10^9$/L	170 - 450 $\times 10^9$/L	

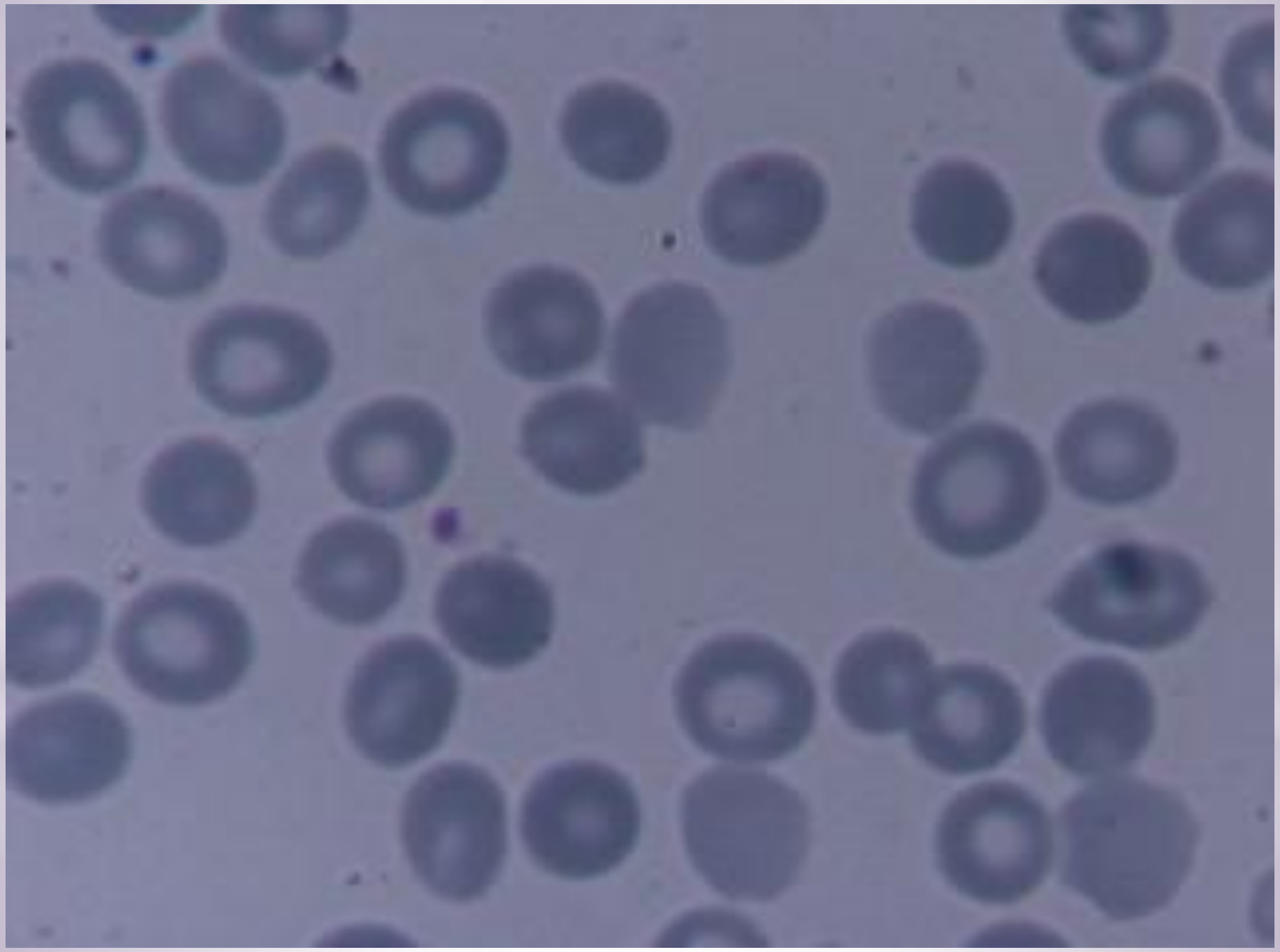
ESR : 36 mm/hr

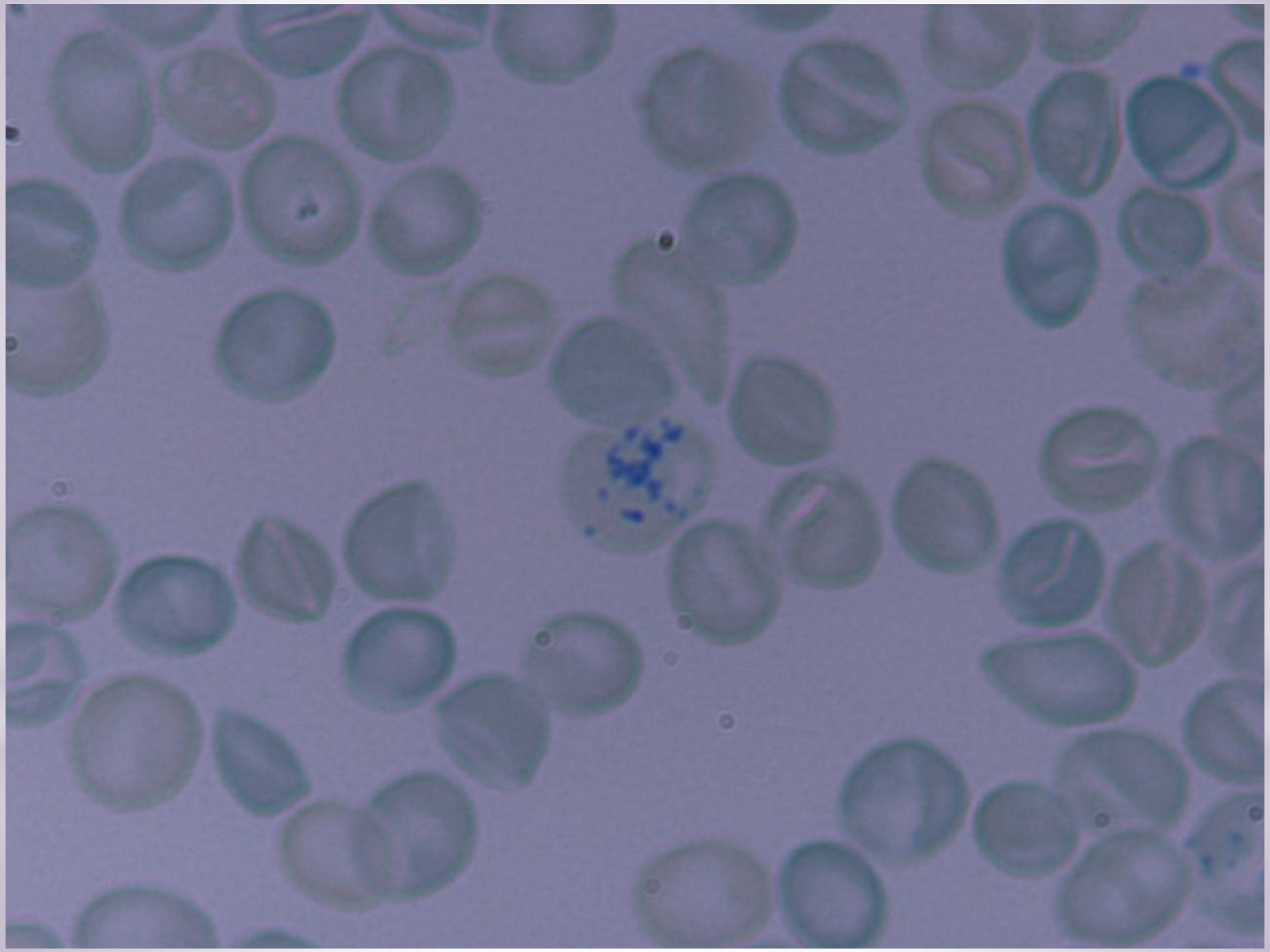
RBC: NORMOCHROMIC NORMOCYTIC RBC with macrocytic RBC.

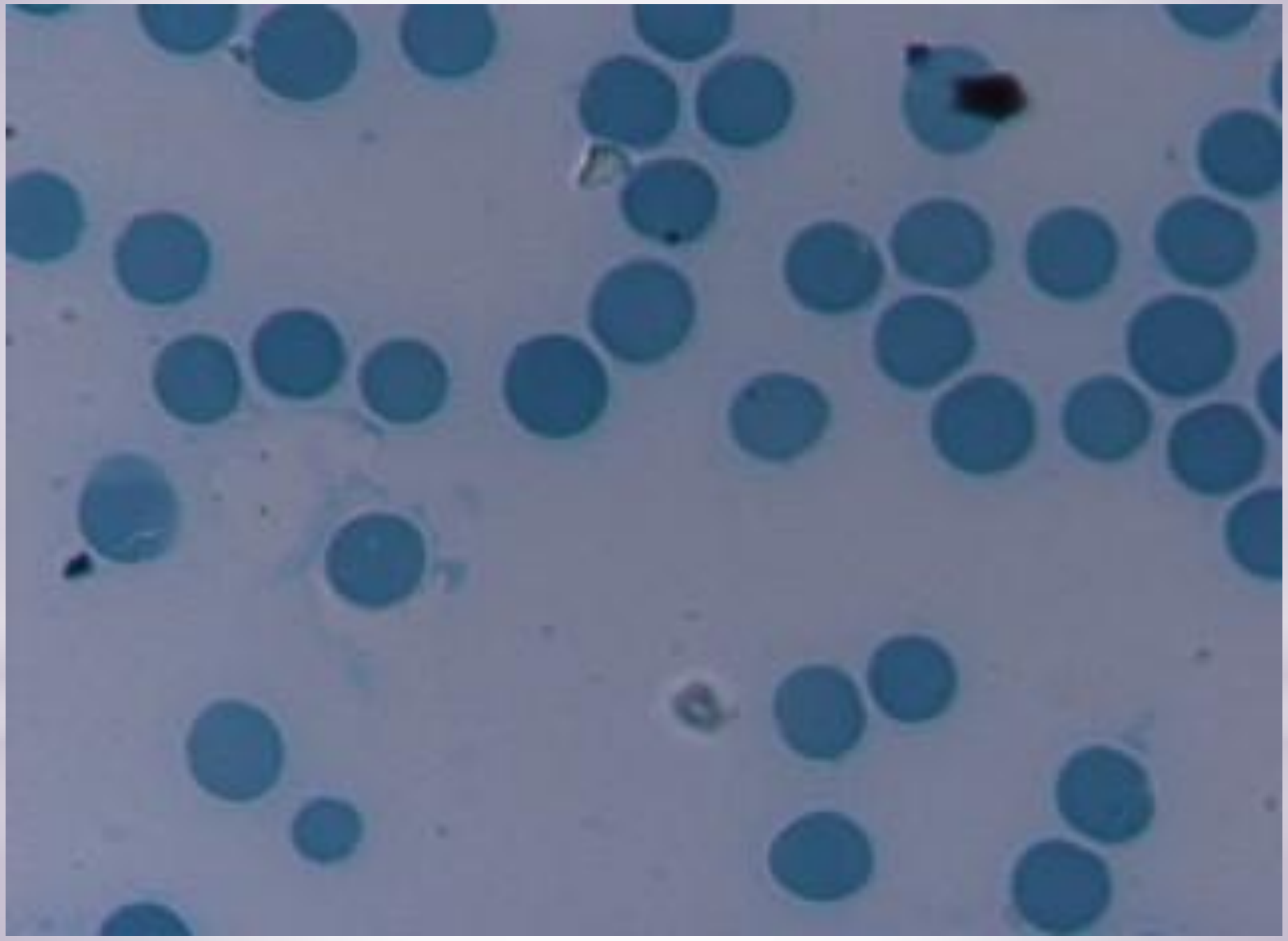
WBC : leukopenia , neutropenia , ALL ARE MATURE.

PLATELET : severely reduced in film.









BONE MARROW ASPIRATE REPORT: (6/1/2013)

Hypocellular bone marrow fragments.

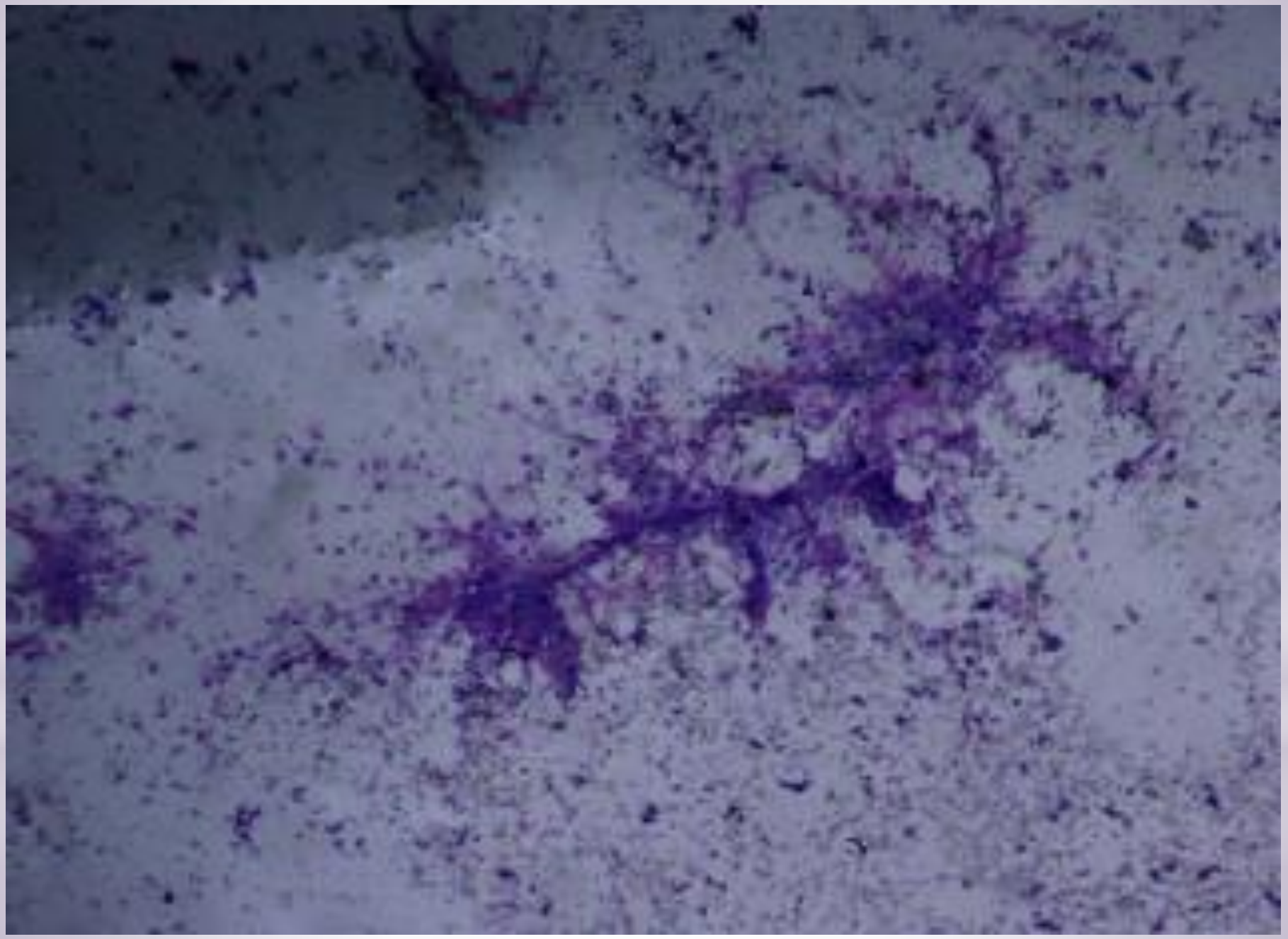
Absent megakaryocytes.

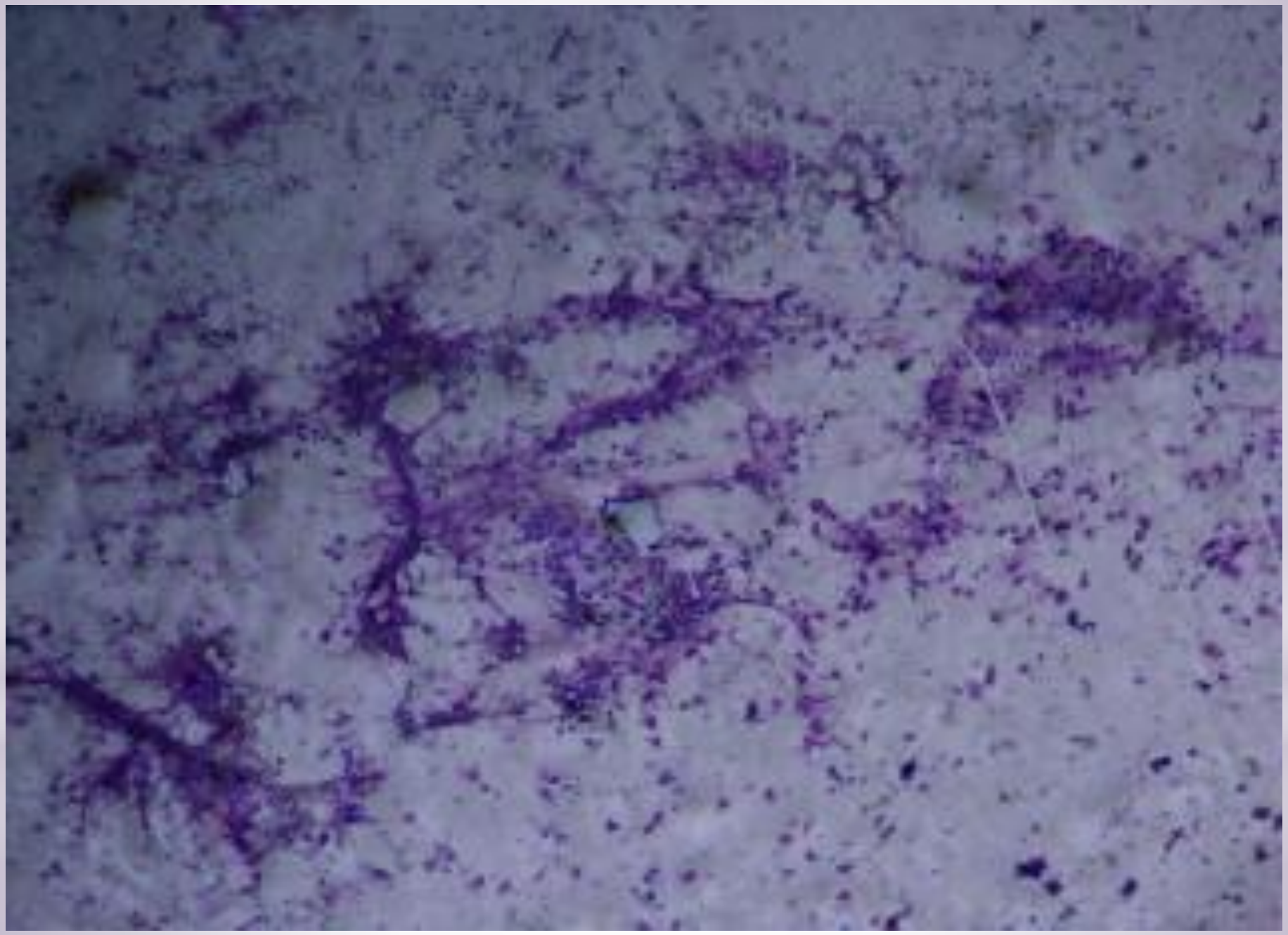
Most of BM fragments shows markedly reduced Hemopoesis .

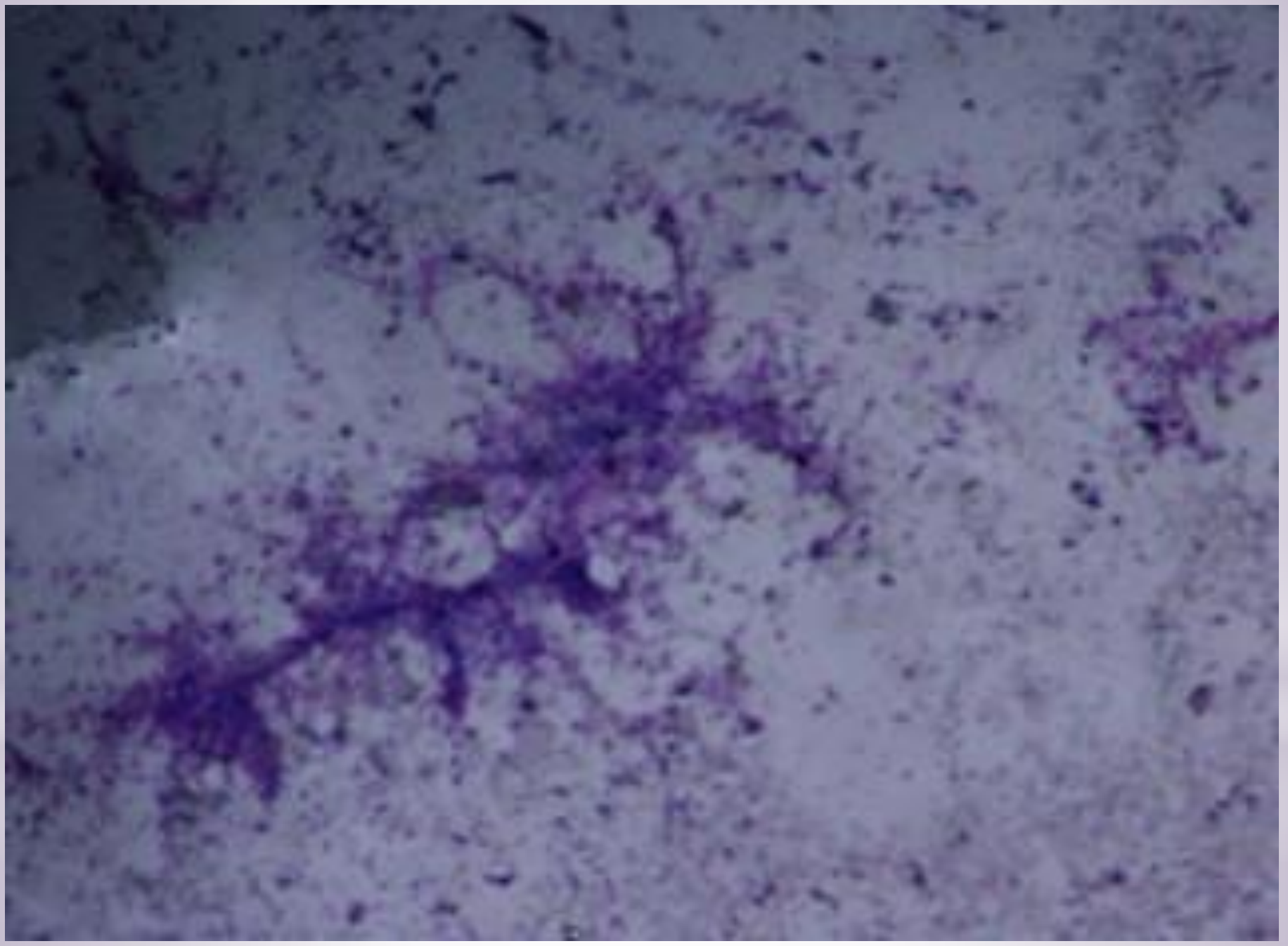
Few of the BM fragments shows mild activity of erythropoiesis (normoblastic) & granulopoiesis (no increase in immature cells)

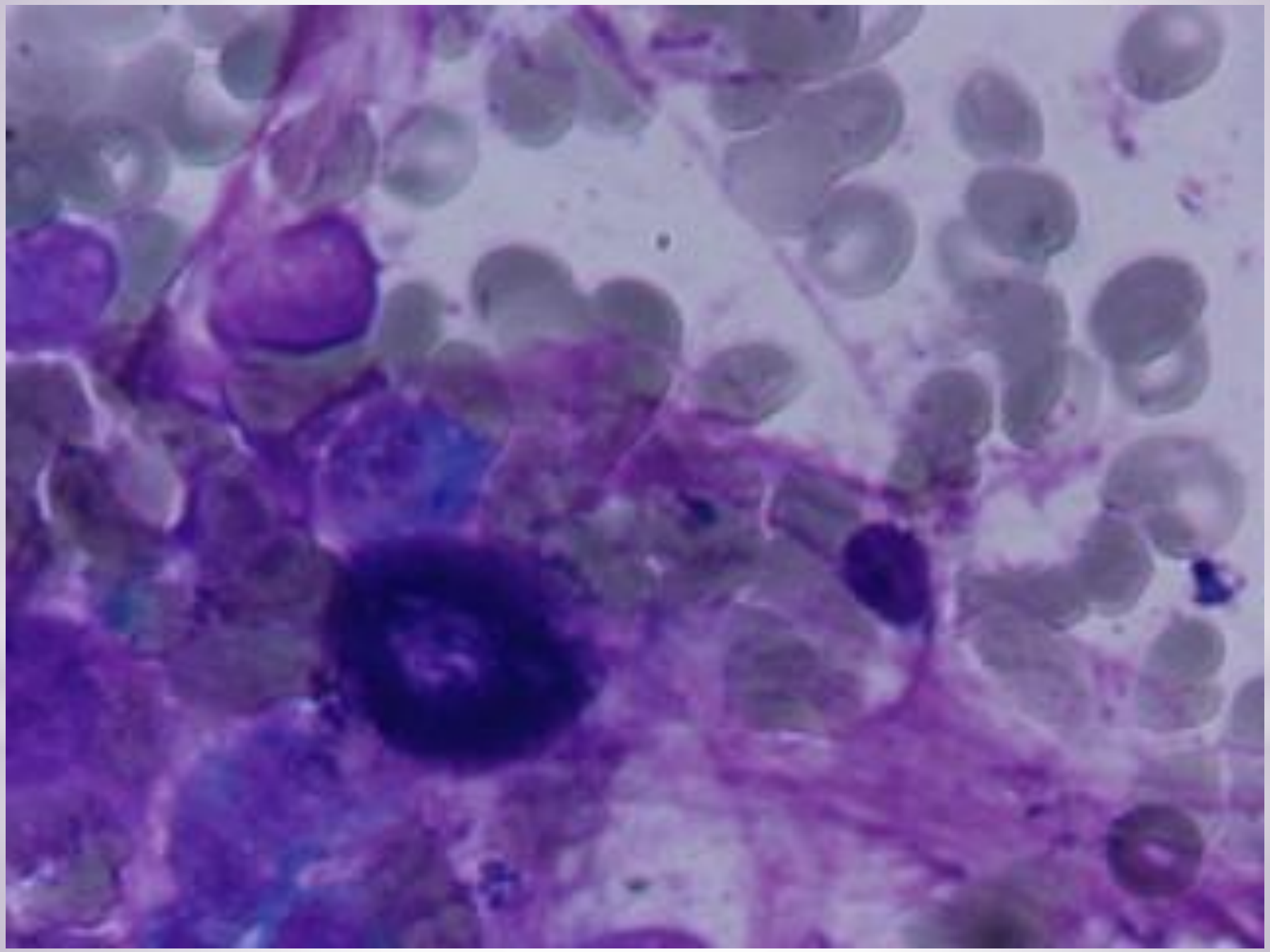
Conclusion :

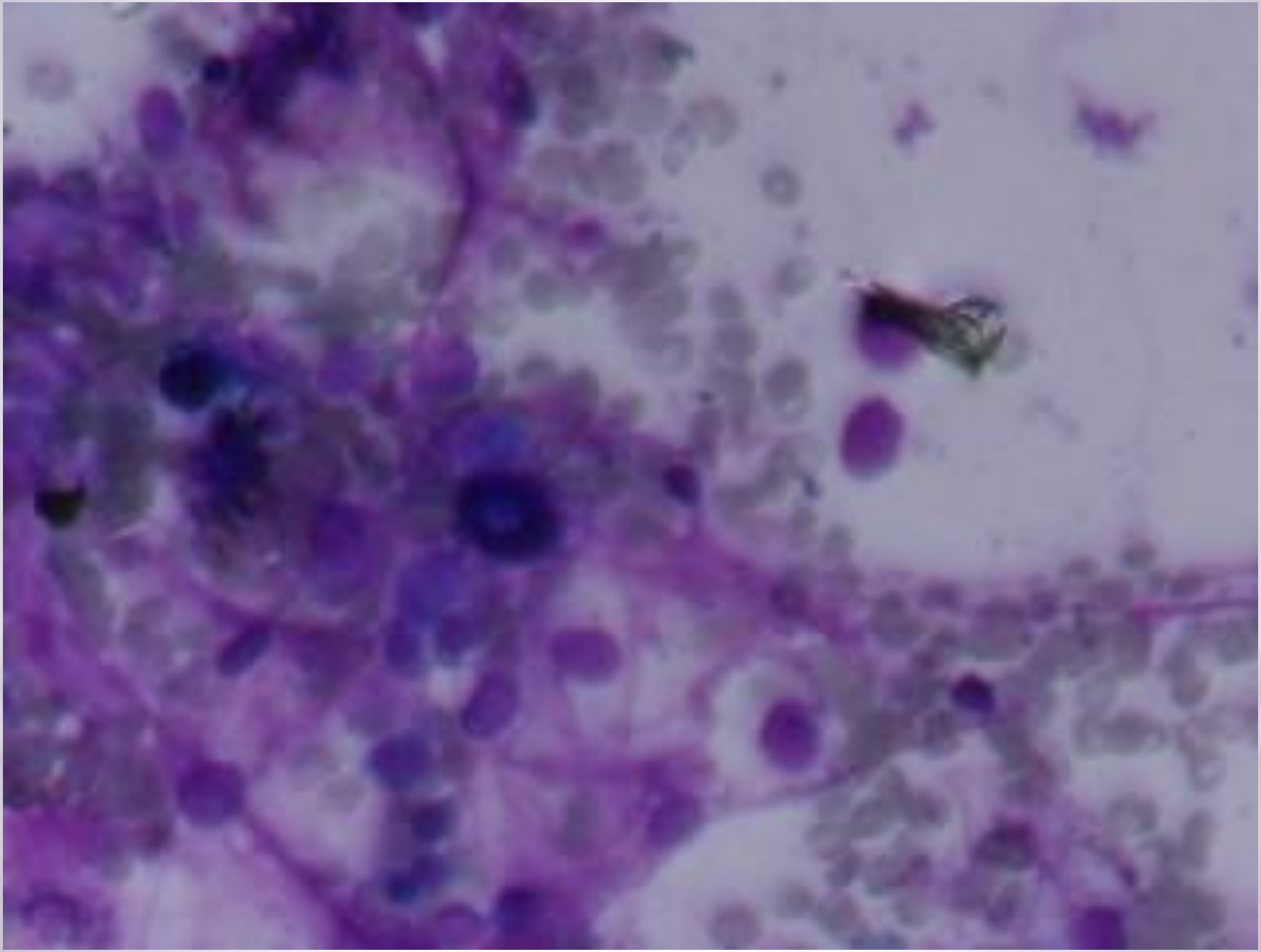
Markedly hypocellular bone marrow











CHROMOSOMAL ANALYSIS OF (HEPARINIZED BLOOD)

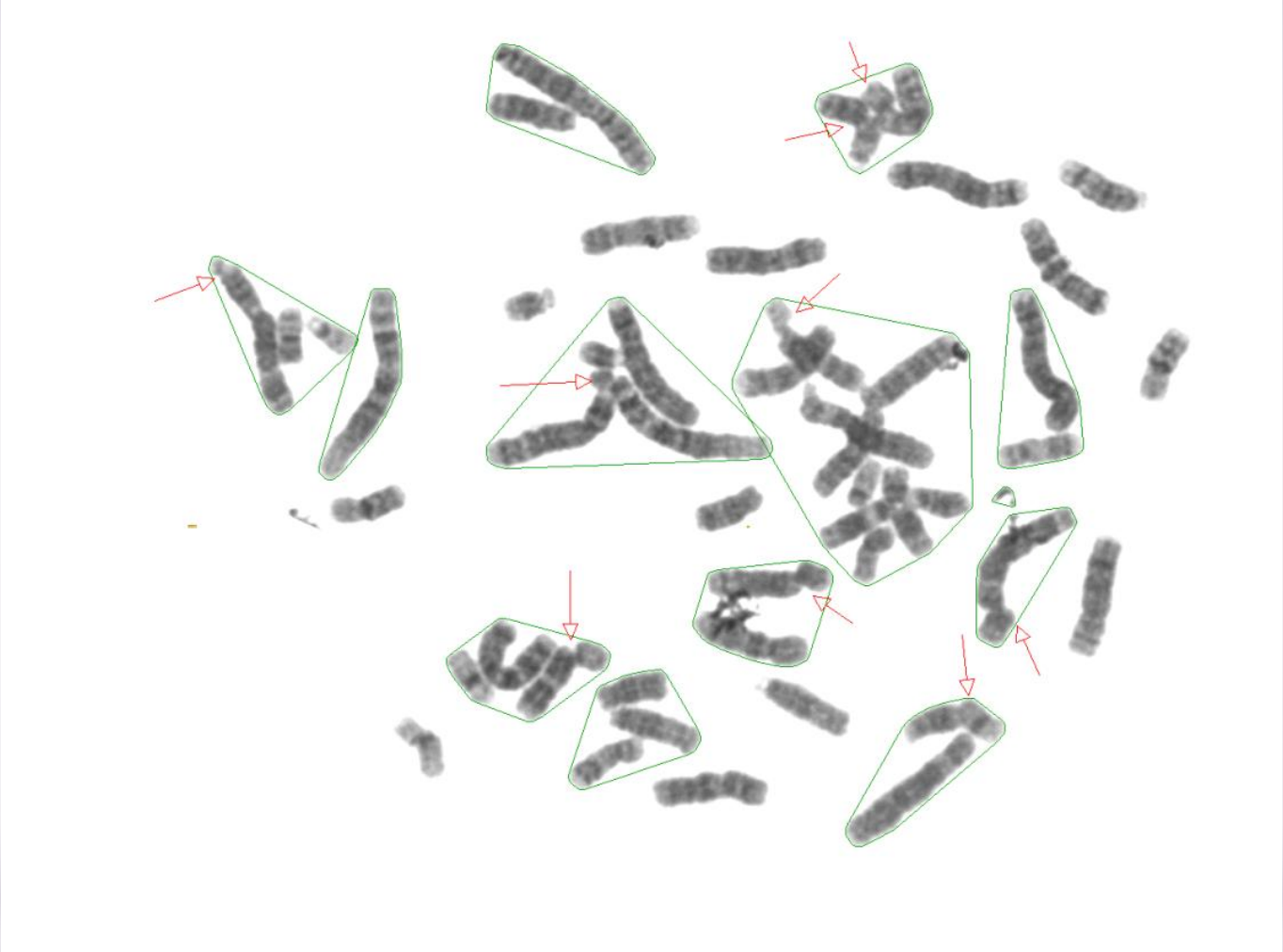
AT : 15 / 2 / 2013

Conclusion : Positive result for spontaneous chromosomal breakage

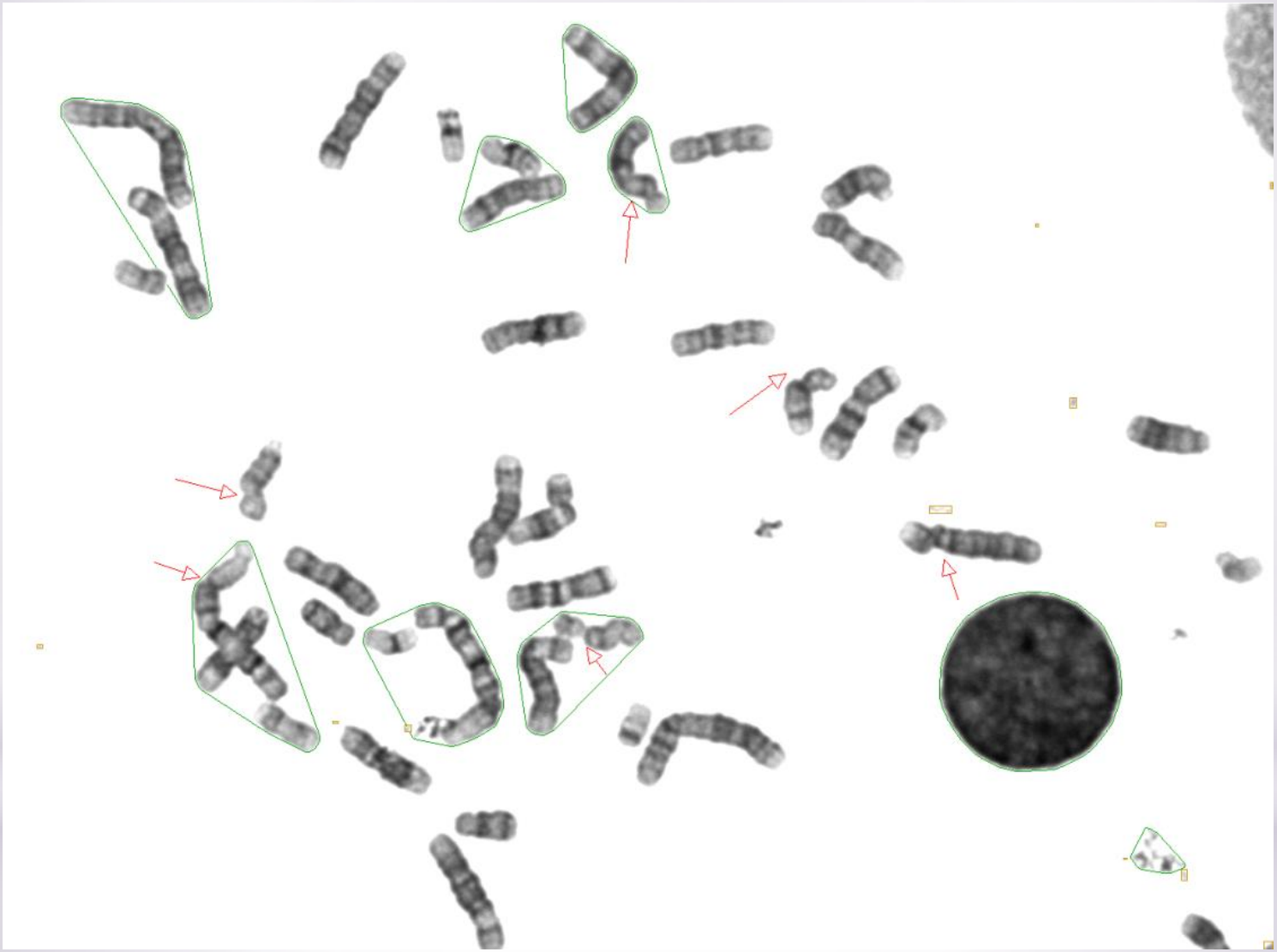
SPONTANEOUS CHROMOSOMAL BREAKAGE TEST PERFORMED ON THIS PATIENT REVEALED A 3.4 BREAKAGE/CELL. THIS IS CONSIDERED A POSITIVE RESULT FOR SPONTANEOUS CHROMOSOMAL BREAKAGE.

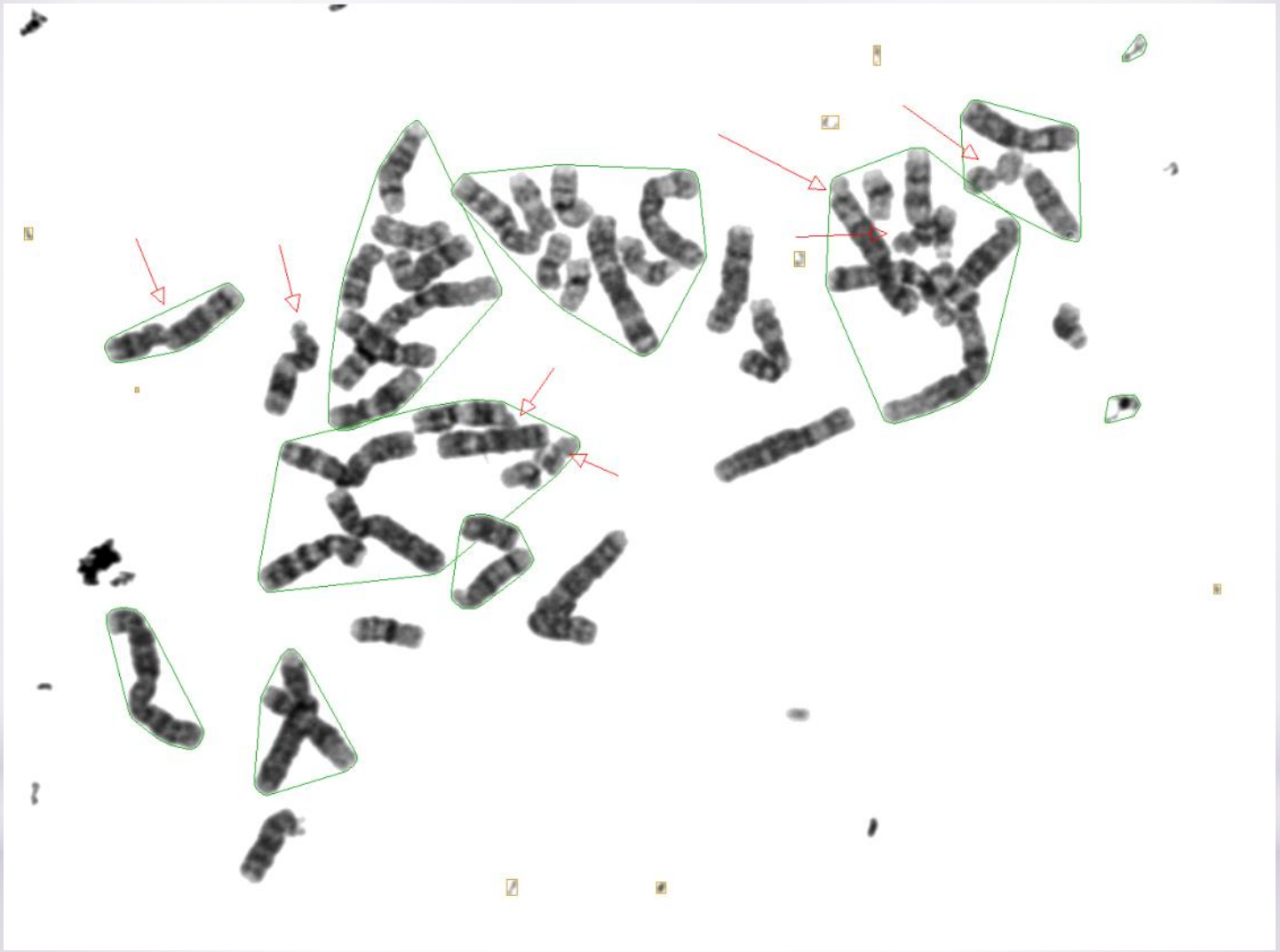
THE DOCUMENTED POSITIVE BREAKAGE LEVEL IS (> 1.0 BREAKS/CELL) .

Cells from FA patients show an abnormally high frequency of spontaneous chromosomal breakage and hypersensitivity to the clastogenic effect of DNA cross-linking agents such as diepoxybutane (DEB) and mitomycin C (MMC). This test is based on the increased chromosomal breakage seen in FA cells compared with normal control subjects after exposure to low concentrations of DEB or MMC ('DEB/MMC stress test').



SPONTANEOUS CHROMOSOMAL BREAKAGE TEST(GTG banding)



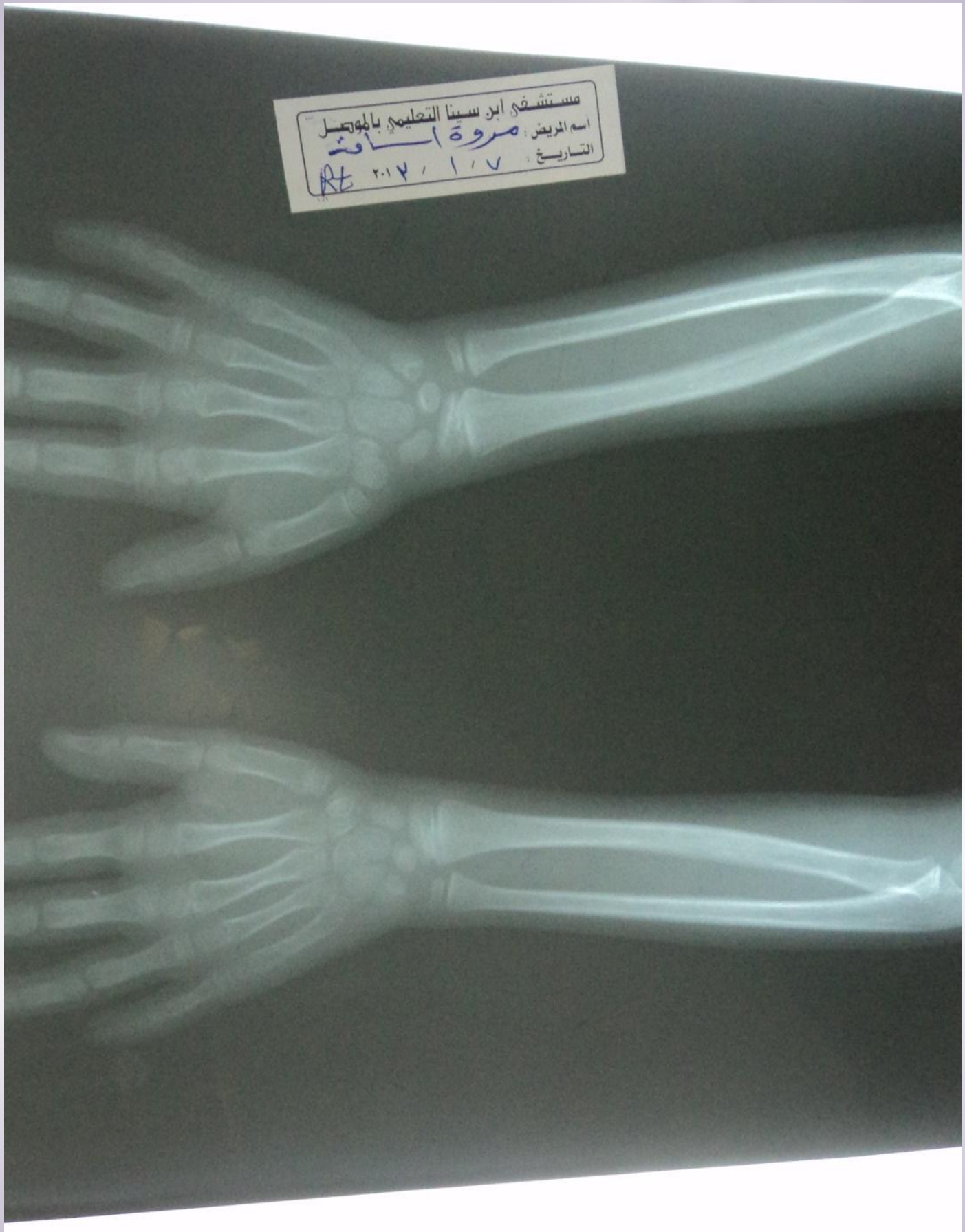


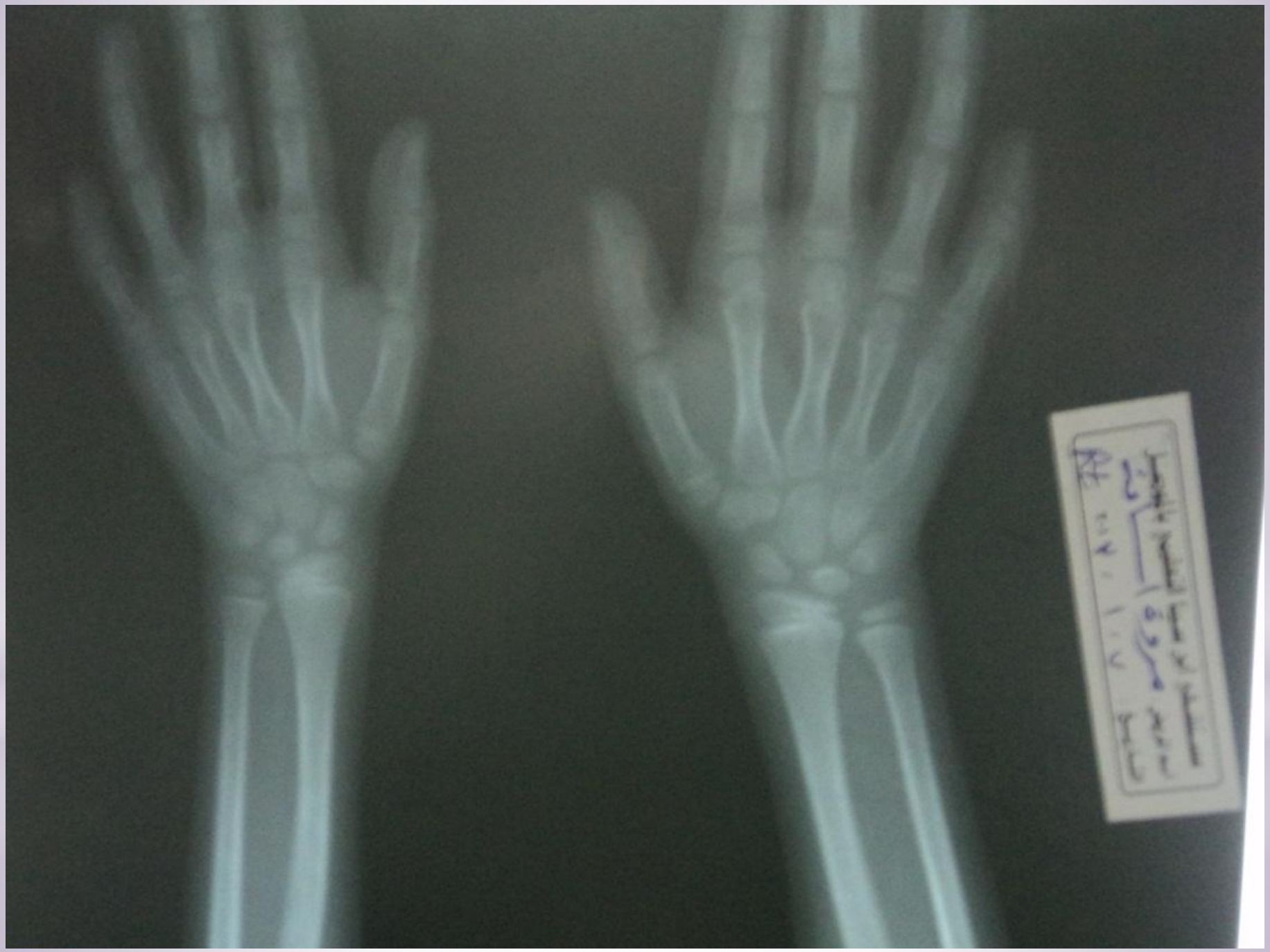
ECG: Normal

U/S AT (7 / 1 / 2013) : Normal

X-RAY (7 / 1 / 2013) : Normal

ECHO AT (7 / 1 / 2013) : Normal


مستشفى ابن سينا التعليمي بالموصل
اسم المريض : مروة اسامة
التاريخ : ٧ / ١ / ٢٠١٢



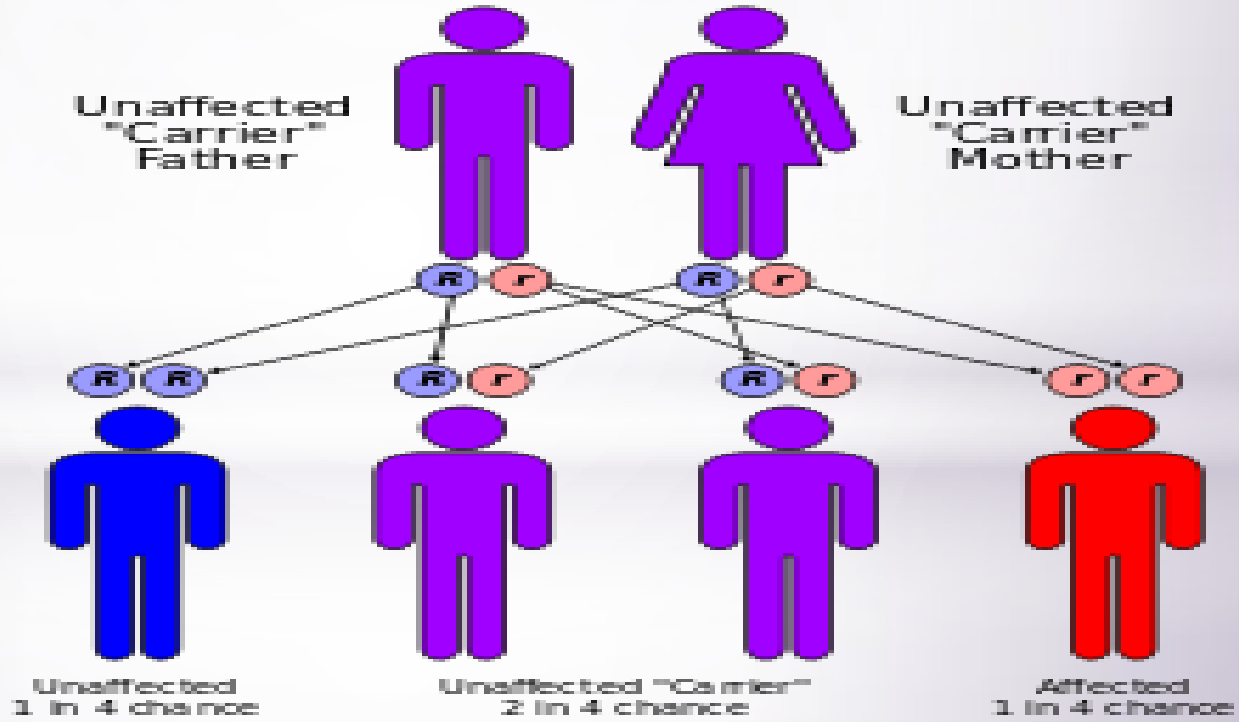
Biochemical tests at : 24 / 2 / 2013

S. Total bilirubin : 10 µmol/L	<17
S. Direct bilirubin : 5 µmol/L	< 5
S. Indirect bilirubin : 5 µmol/L	<12
S. Alkaline phosphatase : 271 U/L	98 – 279
S.A.L.T. : 20 u/L	up to 31
S.A.S.T. : 17 u/L	up to 32
BLOOD UREA: 3.3mmol/L	3.3_7.5
S. Creatinine : 61 µmol/L	53 - 97
S. Uric acid: 275 µmol/L	150_360
S. Ferritin : 54.5 ng/mL	13 – 150
S. Iron : 71 µg/mL	50 – 120
T.I.B.C. : 250 µg/mL	250 - 400


PRINCESS

Fanconi anemia*

Clinical features :FA has become to be recognized as an autosomal recessive disorder in which there is progressive BM failure and an increased **predisposition to malignancy with increasing age** , especially AML (M4,M5) and it may be 1st presentation.



Most, but not all, affected individuals have one or more somatic abnormalities including:

1. skin (café-au-lait spots).
2. skeletal (absent thumbs, radial hypoplasia, scoliosis).
3. genitourinary (underdeveloped gonads, horseshoe kidneys).
4. gastrointestinal.
5. cardiac.
6. neurological anomalies.

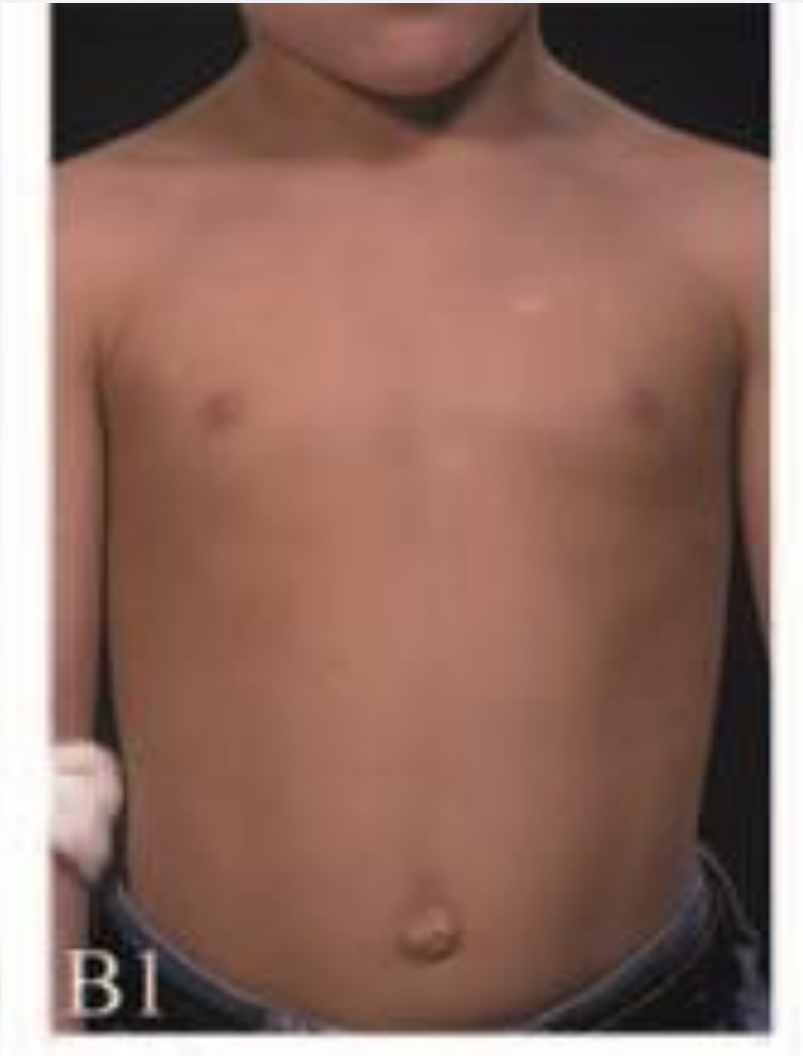
The course of the disease and the pattern of somatic abnormalities show considerable variation , with approximately one-third of patients having no somatic abnormalities. This makes **diagnosis based** on clinical criteria alone difficult and unreliable.

Somatic abnormalities in FA

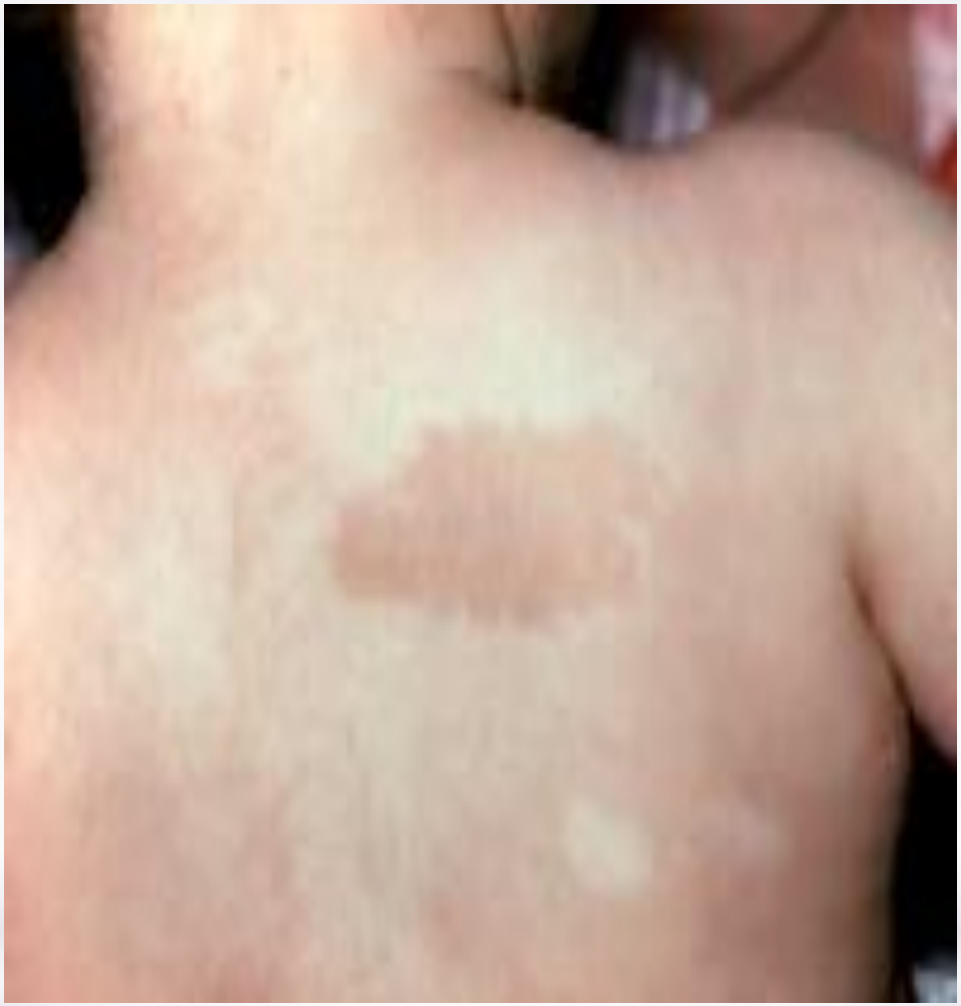
Abnormality	*Percentage of patients*
Skeletal (radial ray, hip, vertebral, scoliosis, rib)	71
Skin pigmentation (café-au-lait, hyper- and hypopigmentation)	64
Short stature	63
Eyes (microphthalmia)	38
Renal and urinary tract	34
Male genital	20
Mental retardation	16
Gastrointestinal (e.g. anorectal, duodenal atresia)	14
Heart	13
Hearing	11
Central nervous system (e.g. hydrocephalus, septum pellucidum)	8
No abnormalities	30

Photographs of FA patients with small mouth and chin (Fanconi faces) *

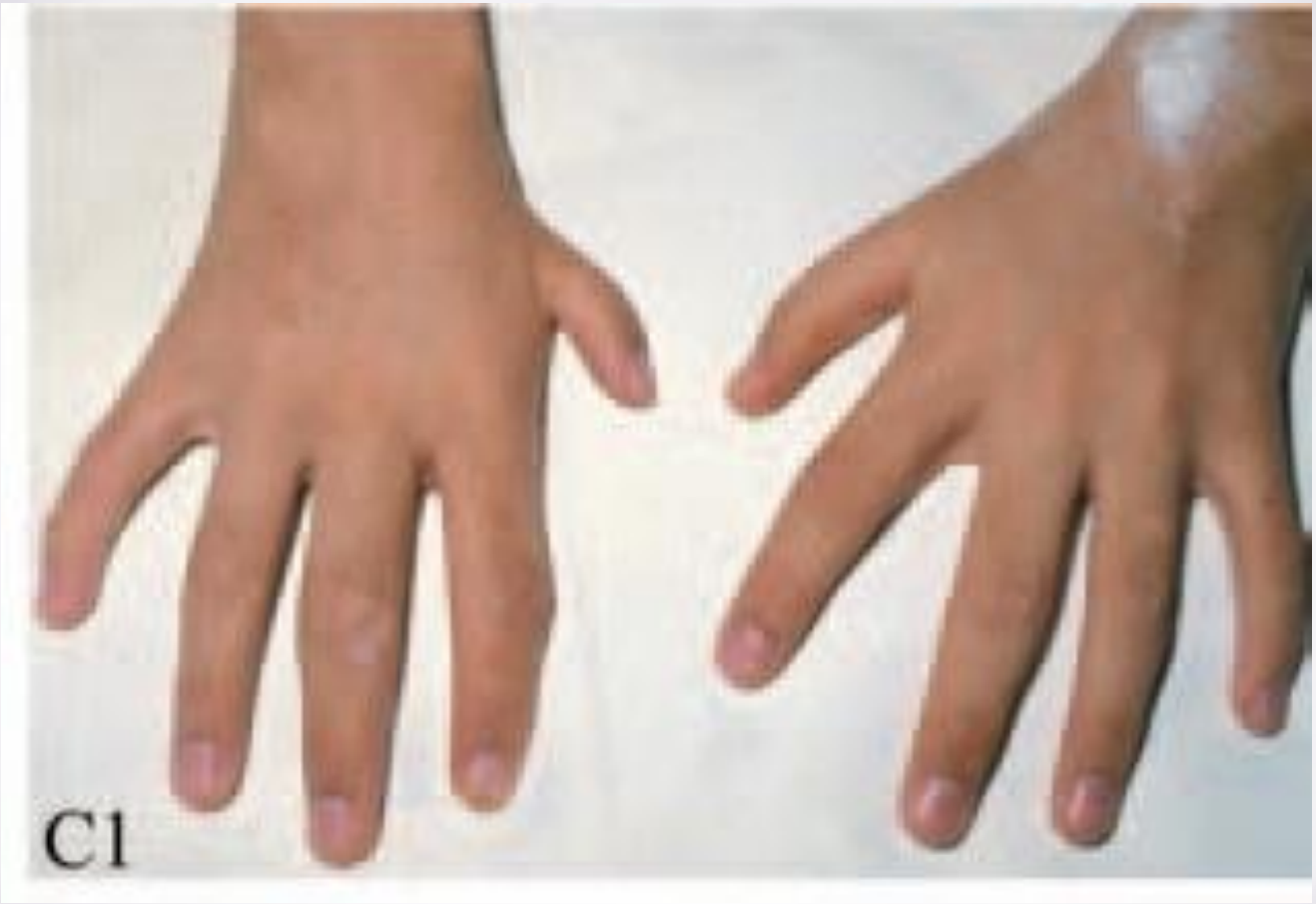
3-year-old patient with Fanconi anemia. Note the multiple birth defects, including short stature, microcephaly, microphthalmia, epicanthal folds, dangling thumbs, site of ureteral reimplantation, congenital dislocated hips, and rocker bottom feet *



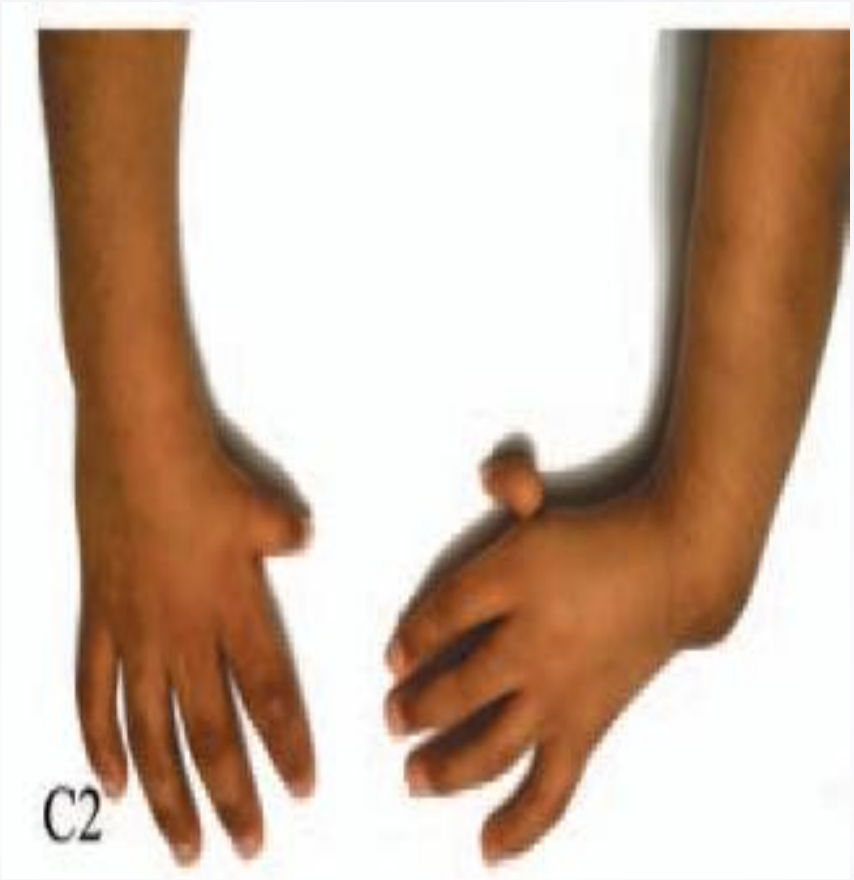
Abnormalities of pigmentation * (hyper- and hypopigmentation) on the abdomen



café-au-lait spot and a hypopigmented patch *



Hands/forearms of FA children showing * hypoplastic thumbs



rudimentary (‘dangling’) thumbs *

At birth the blood count is usually normal. Pancytopenia presents in most cases between the ages of 5 and 10 years (median age 7 years). However, in some cases the pancytopenia develops in adolescence or even in adult life.

The (Hb) and platelet count **are usually first to fall ;** the granulocytes are usually well preserved in the early stages.

As the pancytopenia develops, the BM becomes progressively hypocellular.There is often a marked increase in macrophage activity with evidence of haemophagocytosis.

BM failure leading to fatal hemorrhage or infection which is the main cause of death in FA patients.

In a recent analysis from the International Fanconi Anemia Registry (IFAR), the median survival time was 24 years.

The cumulative incidence of hematological malignancy by the age of 40 years is 33%. Besides these hematological malignancies, there is a significant risk of hepatic tumors and squamous cell carcinoma, including squamous cell carcinomas of the vulva, esophagus, head and neck. The cumulative incidence of solid tumors is calculated to be 28% by the age of 40 years.

The impression is that malignancies occur mainly in patients with late-onset BM failure and longer survival, with a median age of 13 years for leukemia and 25 years for solid tumors. Furthermore, long-term follow-up in FA patients who have been treated by haemopoietic stem cell transplantation (SCT) is showing a higher incidence of non-haematological malignancies in patients with FA than patients with other types of BM failure who underwent SCT, again emphasizing the predisposition to malignancy.

thanks