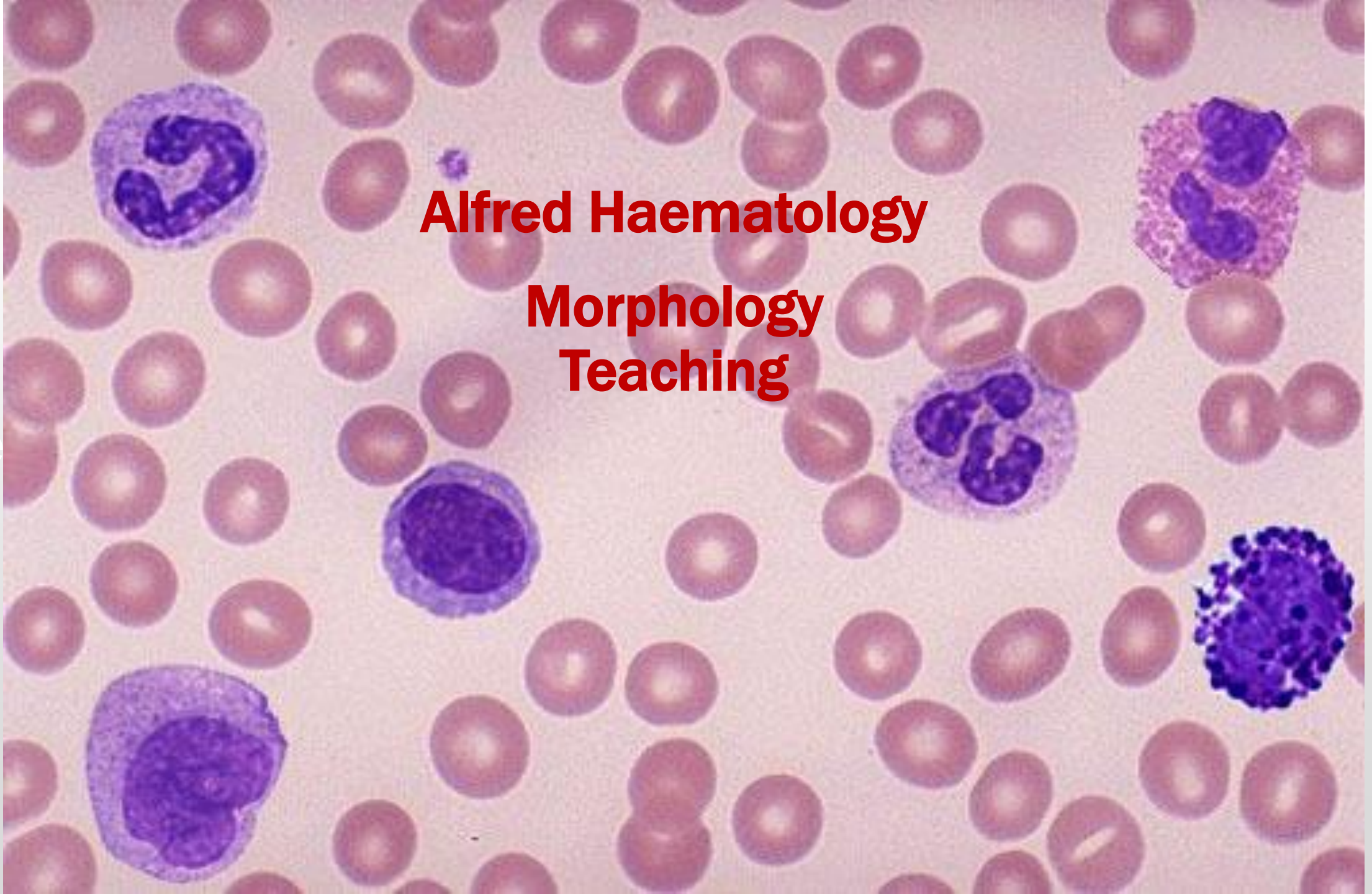


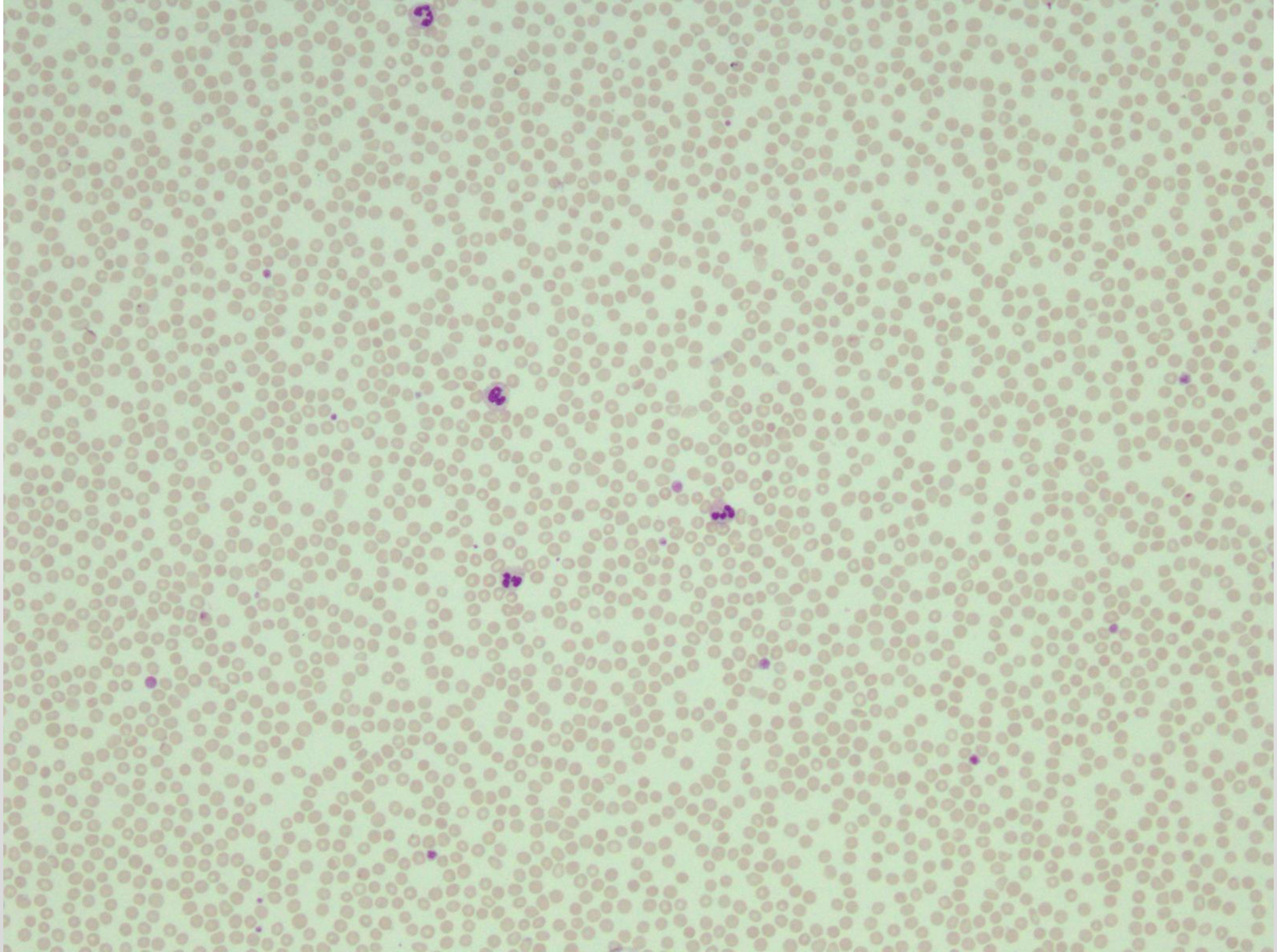
Title

Case 1: A family with low platelets

- 29 yo man

- Receiving regular IVIg for known congenital immunodeficiency
- Father and brother have thrombocytopenia
- Mother has Ehlers-Danlos syndrome

- No abnormal bleeding

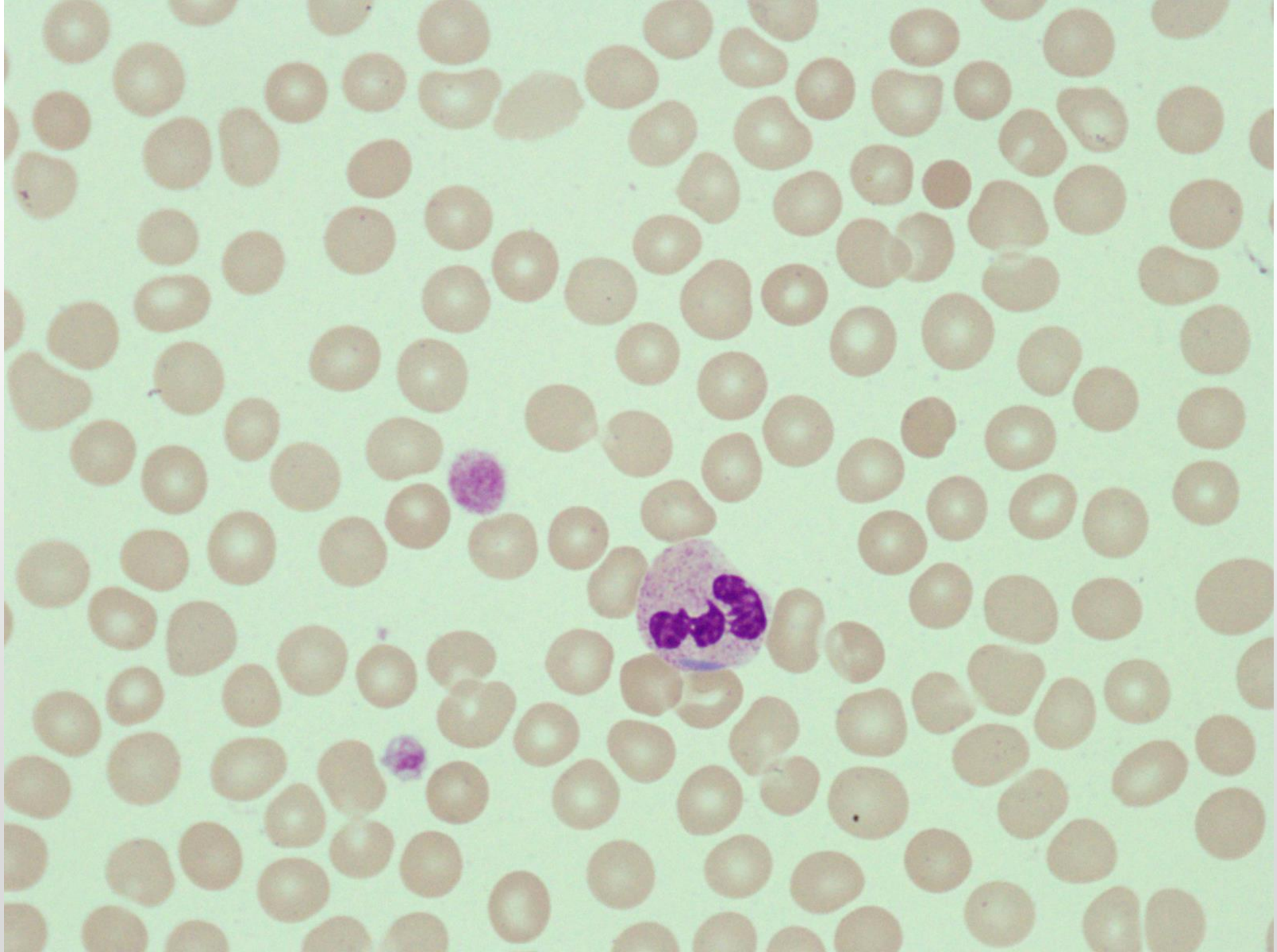


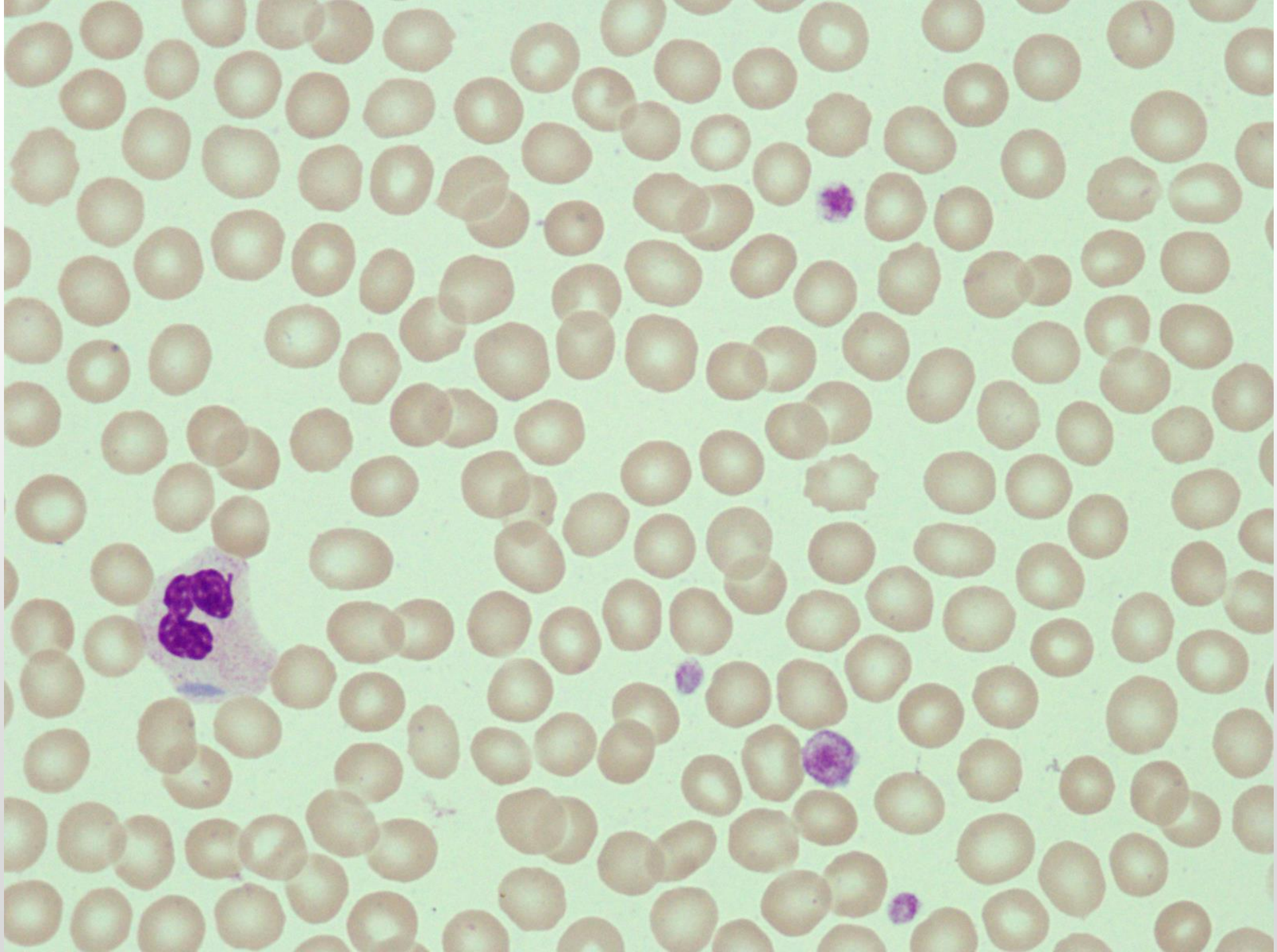
Hb 158 g/L

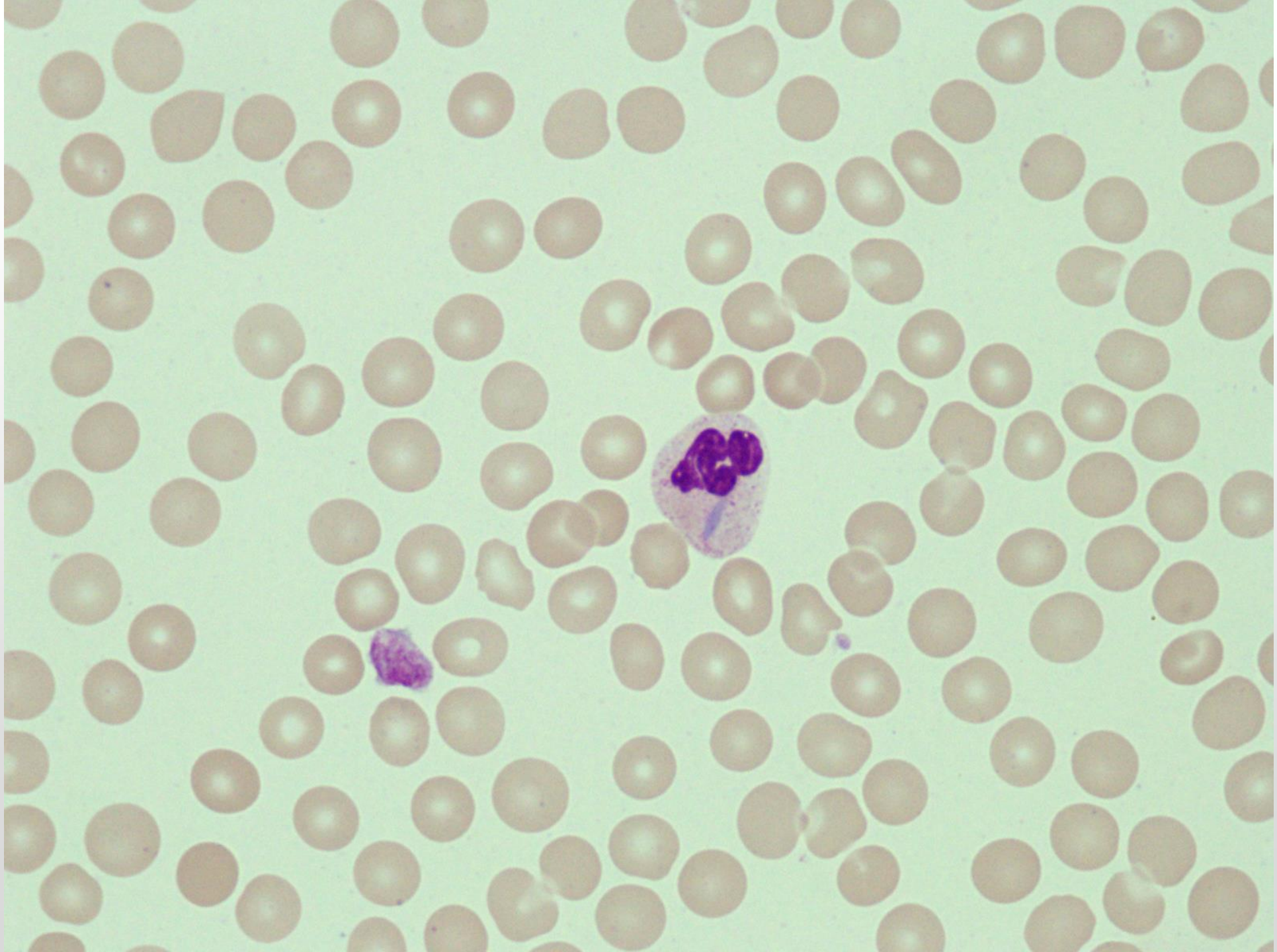
WCC 2.7 x10^9/L

PLT 14 x10^9/L

Can you pick out the platelets?

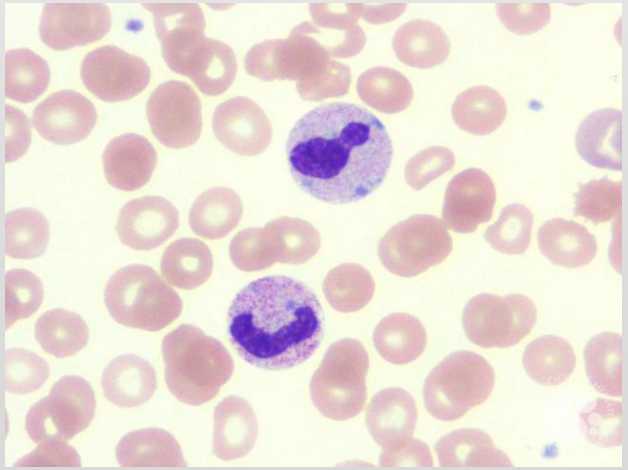






Final Diagnosis: May-Hegglin anomaly

- Morphology: macrothrombocytopenia AND Döhle body-like inclusions
- Döhle body-like inclusions: seen throughout cell, not just at edge; spindle- or crescent-shaped; composed of mutant non-muscle myosin heavy chain protein (*MYH*9)
- Compare with 'ordinary' Döhle bodies: composed of ribosomes and glycogen, seen in sepsis, dysplasia



- Autosomal dominant
- Reduced glycoprotein 1b/IX/V complex

Case 2: A tale of two sisters

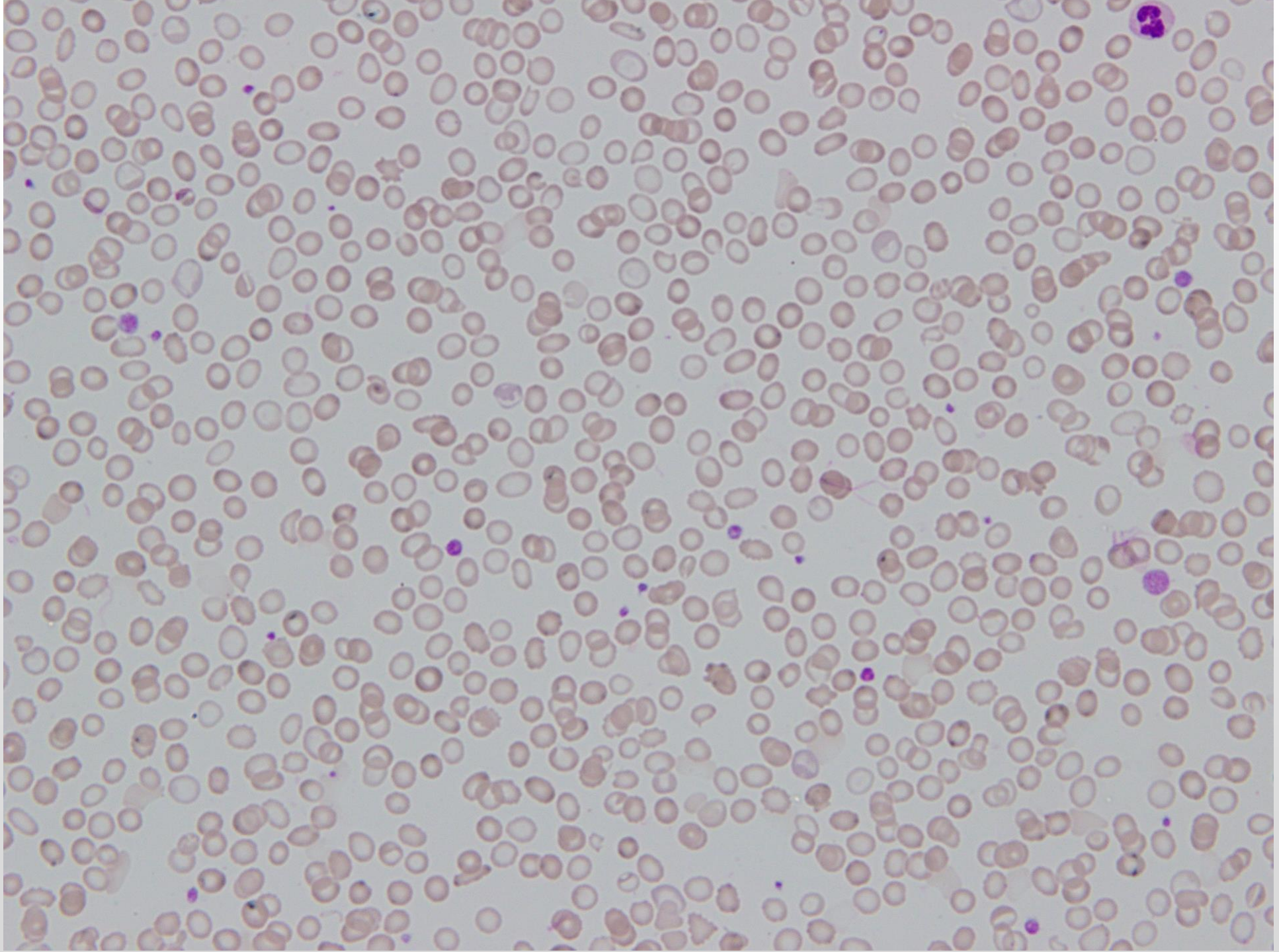
MS: 19 yo

- Recurrent epistaxis
- Menorrhagia
- Frequent iron infusions for anaemia

- Parents are second cousins

TS: 22 yo

- Iron deficient in past
- Wants to have a baby

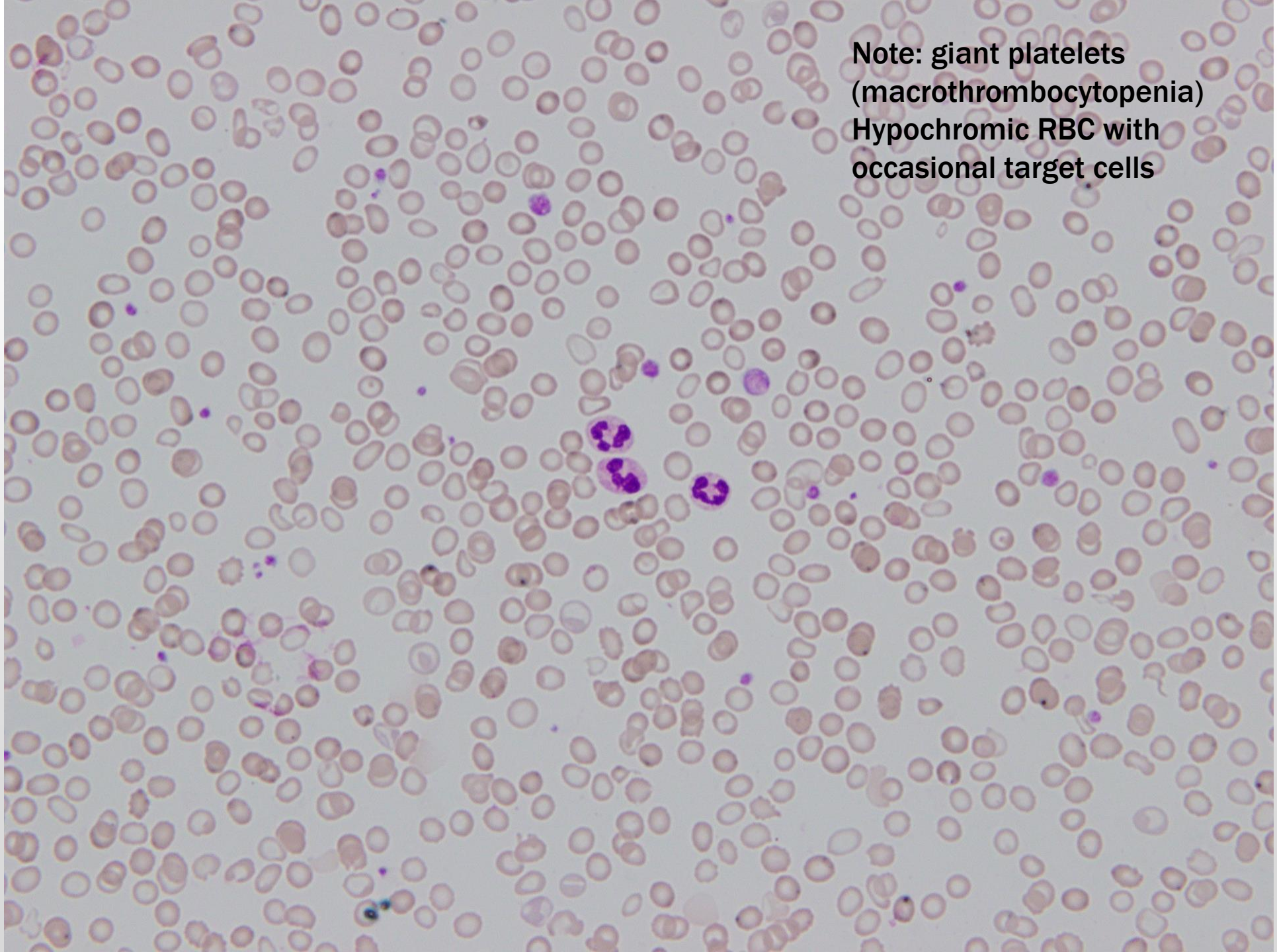


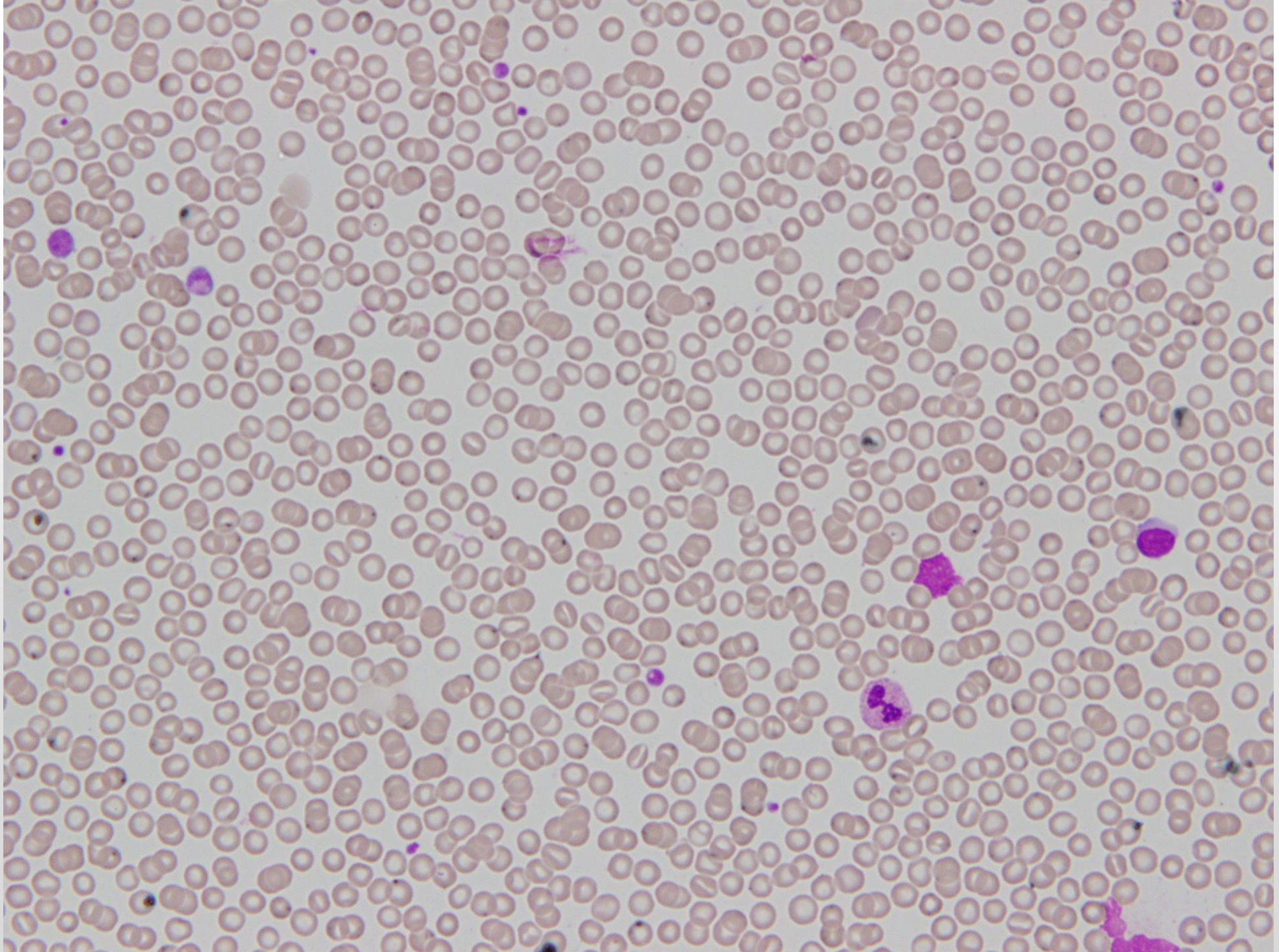
Mrs MS

Hb 67 g/L
(MCV 79 fL, MCH 23.5 pg)

WCC 4.97 x 10^9/L

Platelets ???


Note: giant platelets
(macrothrombocytopenia)
Hypochromic RBC with
occasional target cells

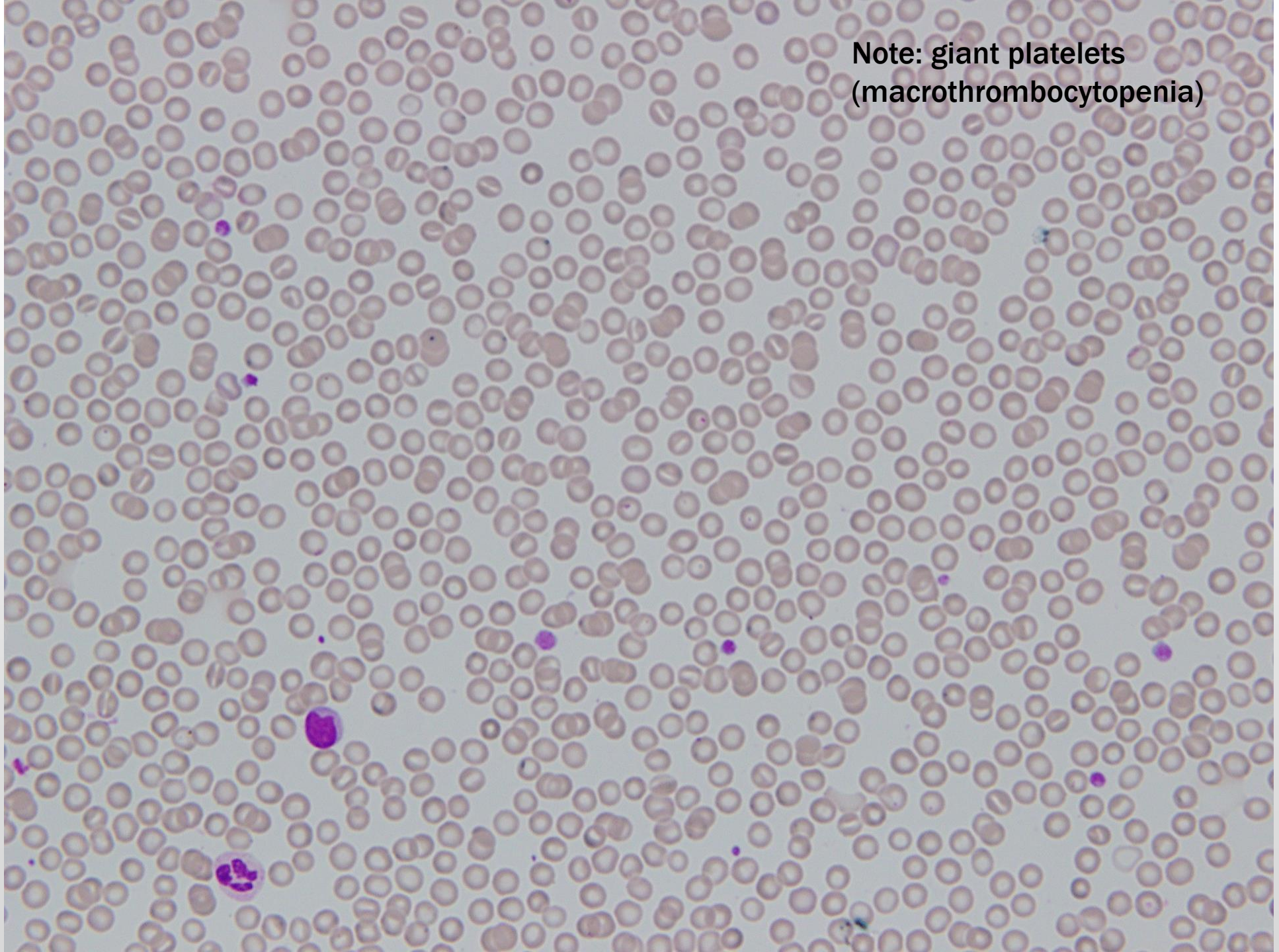


Miss TS

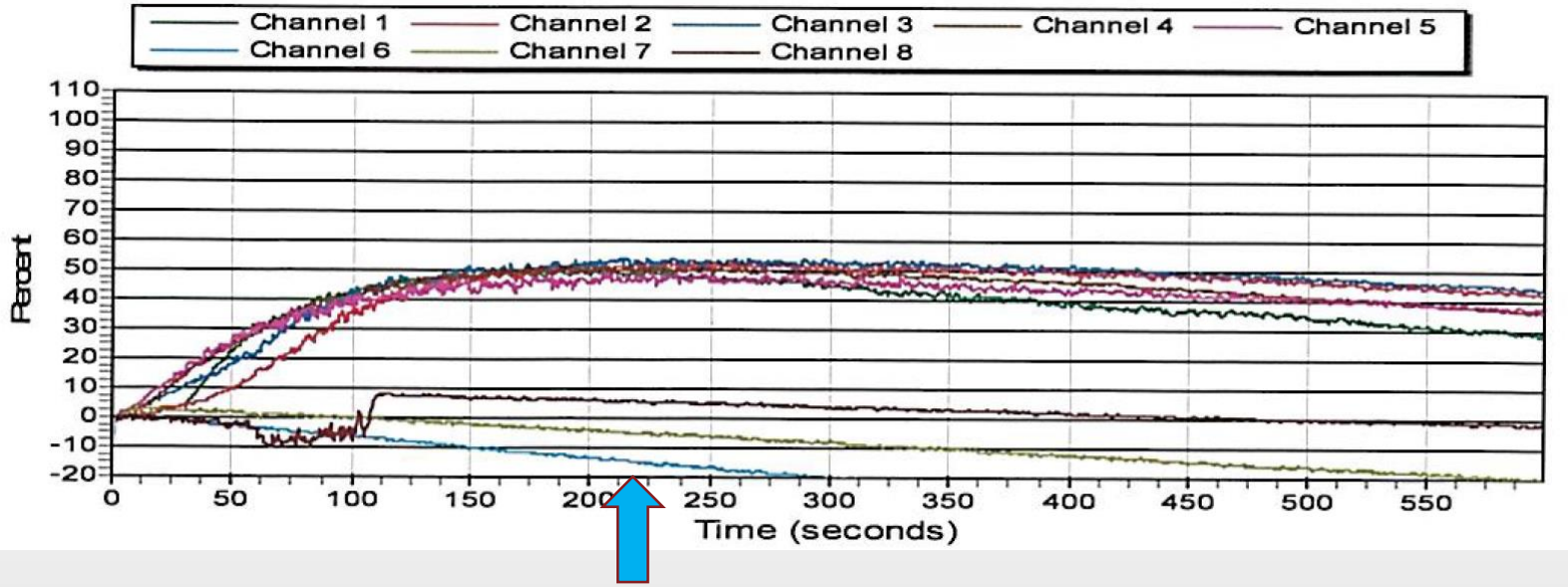
Hb 146 g/L
(MCV 93 fL, MCH 31 pg)

WCC 10.24 x 10^9/L

Platelets ???

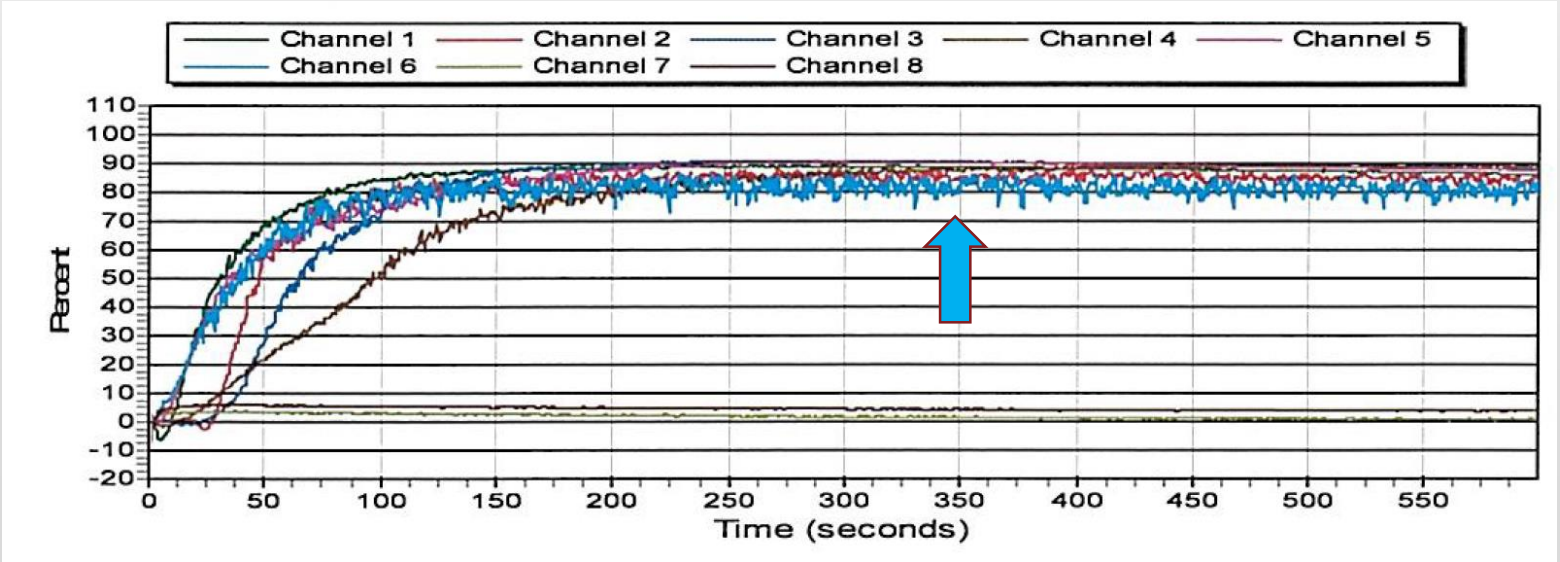

Note: giant platelets
(macrothrombocytopenia)

Platelet aggregation results



Miss TS

Absent aggregation with high dose ristocetin (blue)

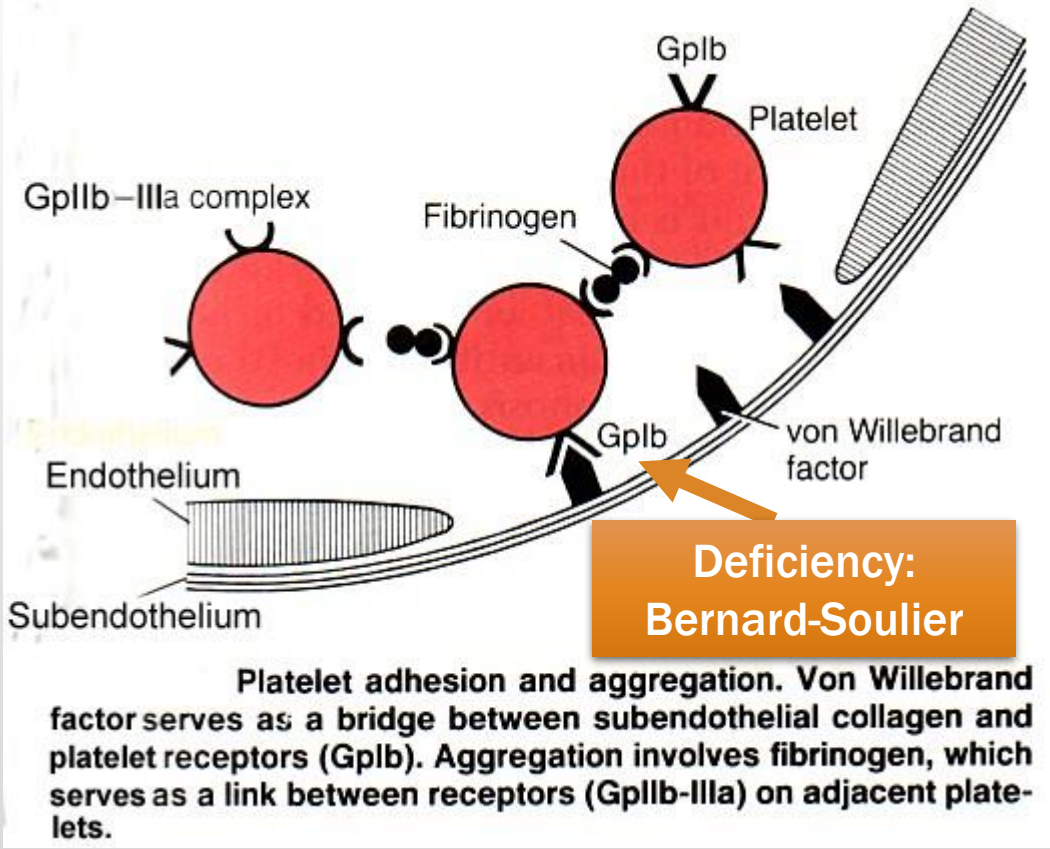


Normal patient

Normal aggregation with high dose ristocetin (blue)

Diagnosis: Bernard Soulier syndrome

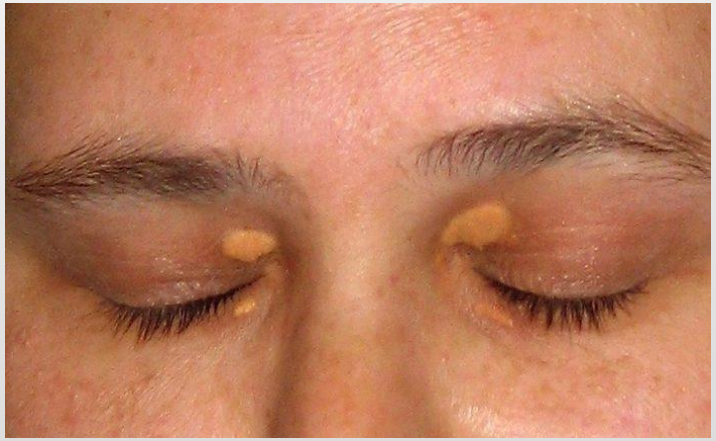
- Autosomal recessive
- Mutations in GP-1b or GP-IX
- Causes abnormalities in glycoprotein 1b/IX/V complex, leading to defect in vWF-dependent platelet aggregation



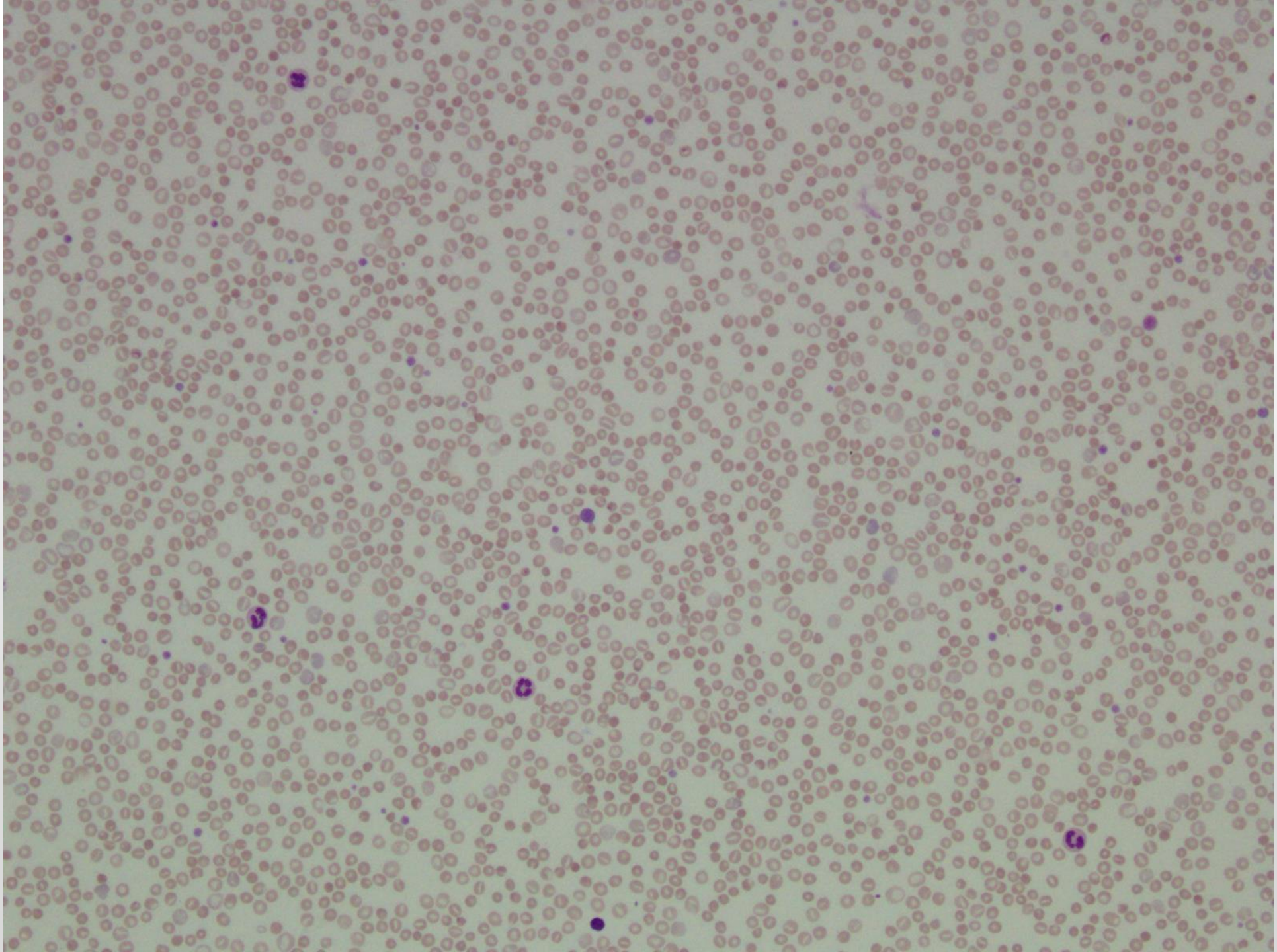
Platelet adhesion and aggregation. Von Willebrand factor serves as a bridge between subendothelial collagen and platelet receptors (GpIb). Aggregation involves fibrinogen, which serves as a link between receptors (GpIIb-IIIa) on adjacent platelets.

Case 3: The importance of morphology

- 61 yo man, Croatian ancestry
- 30 year history of “ITP”
- Recurrent iron deficiency anaemia
- Periorbital xanthomas



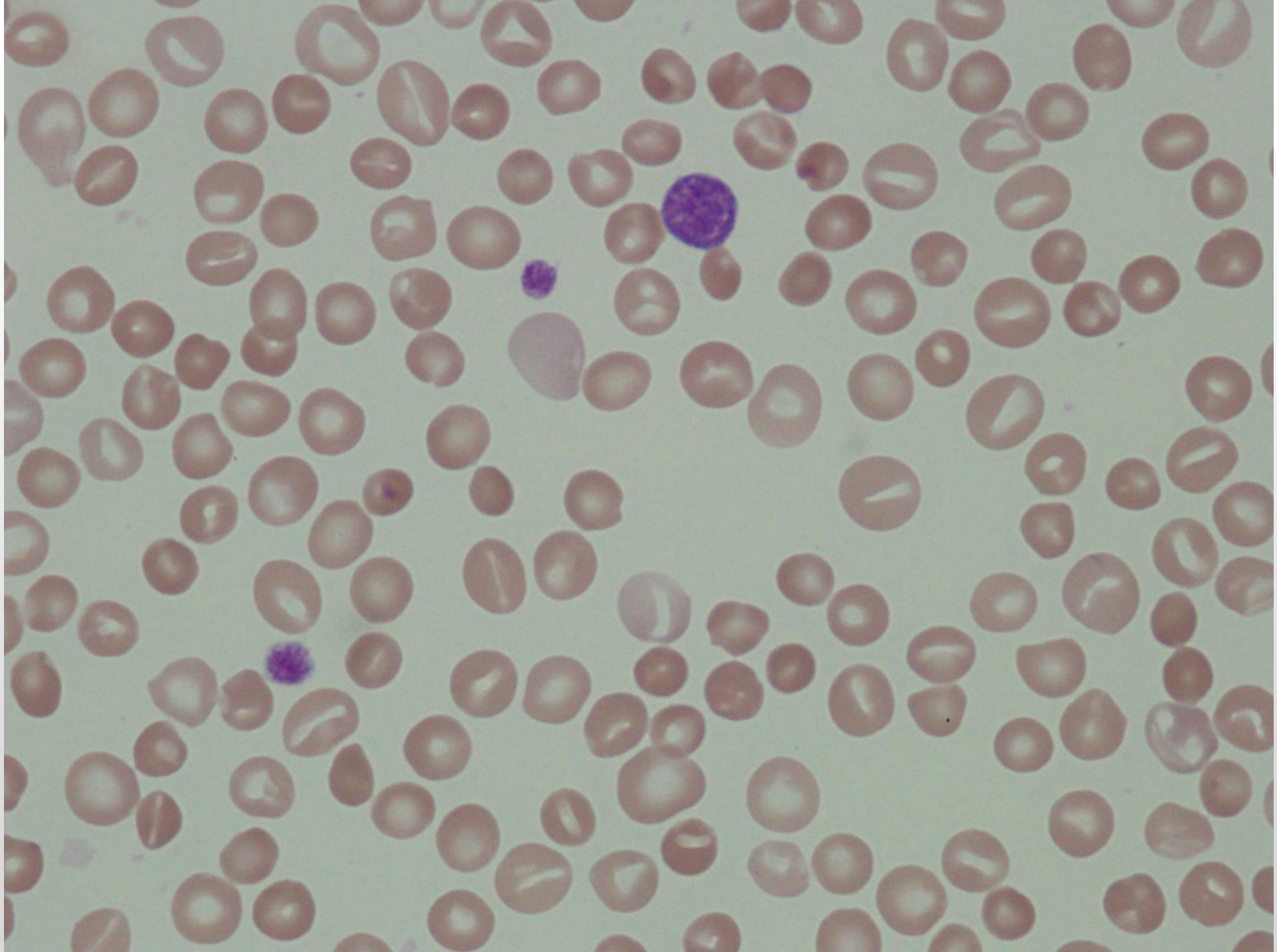
Xanthomas: NOT this patient!

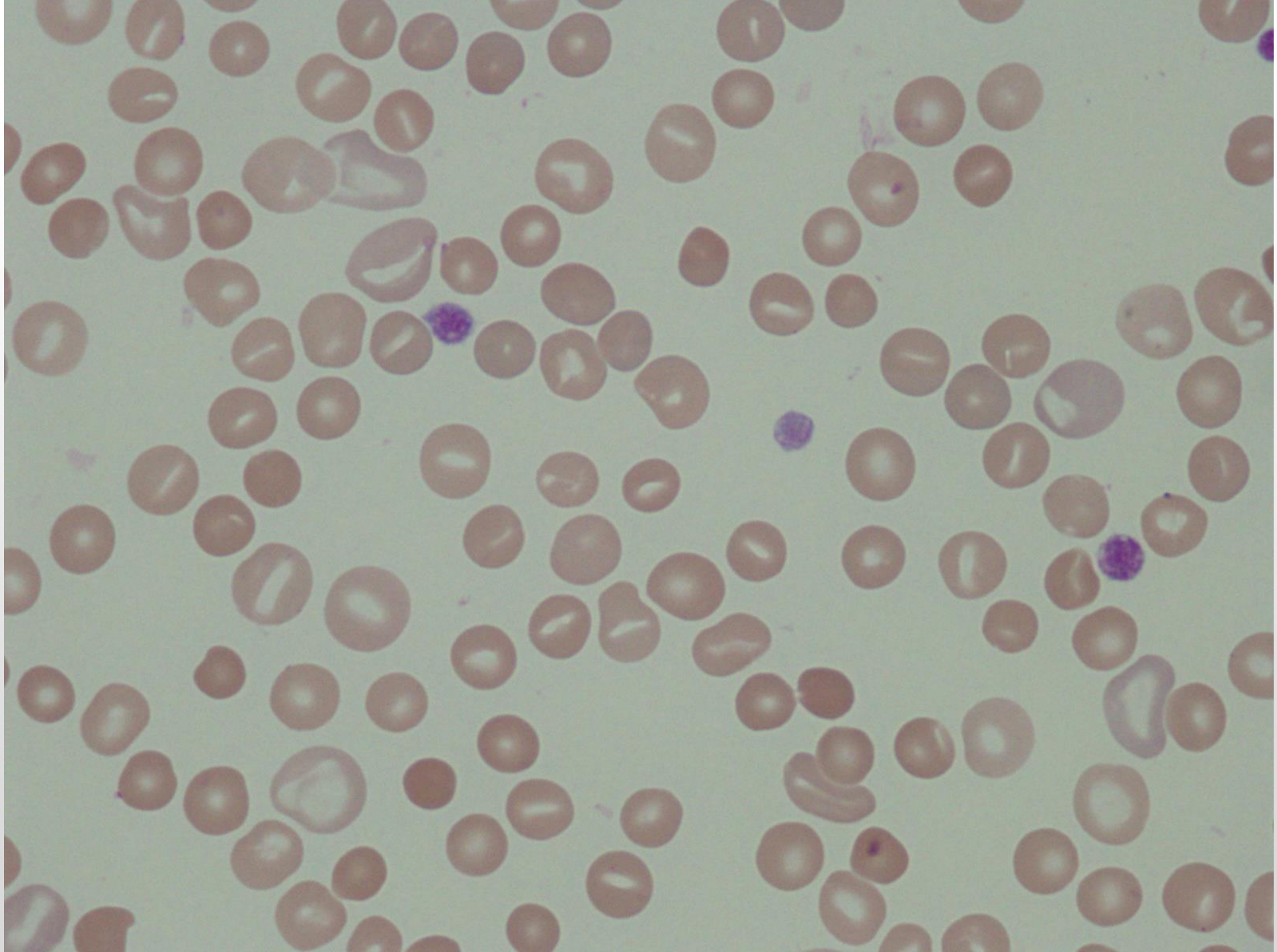


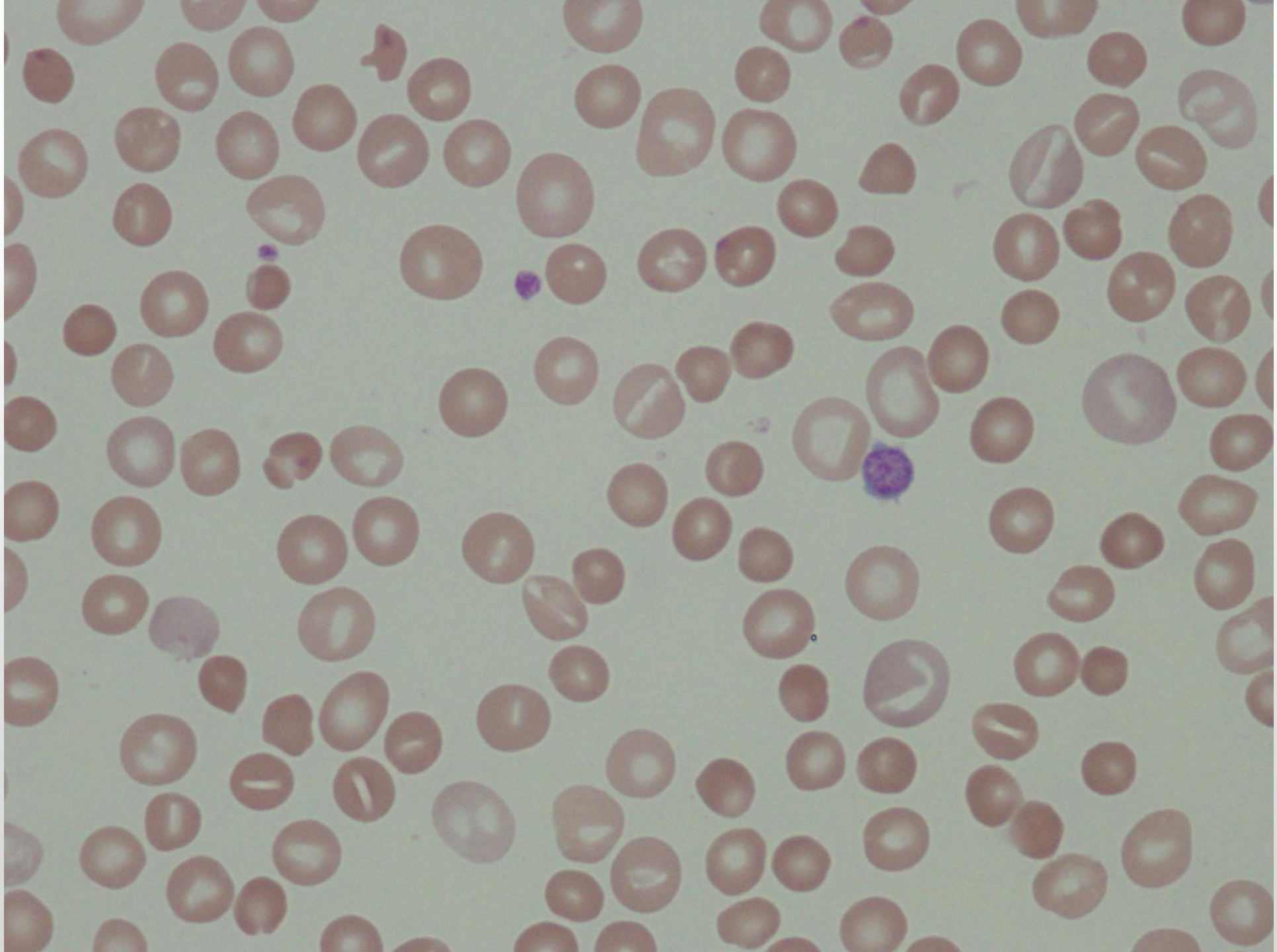
Hb 86 g/L

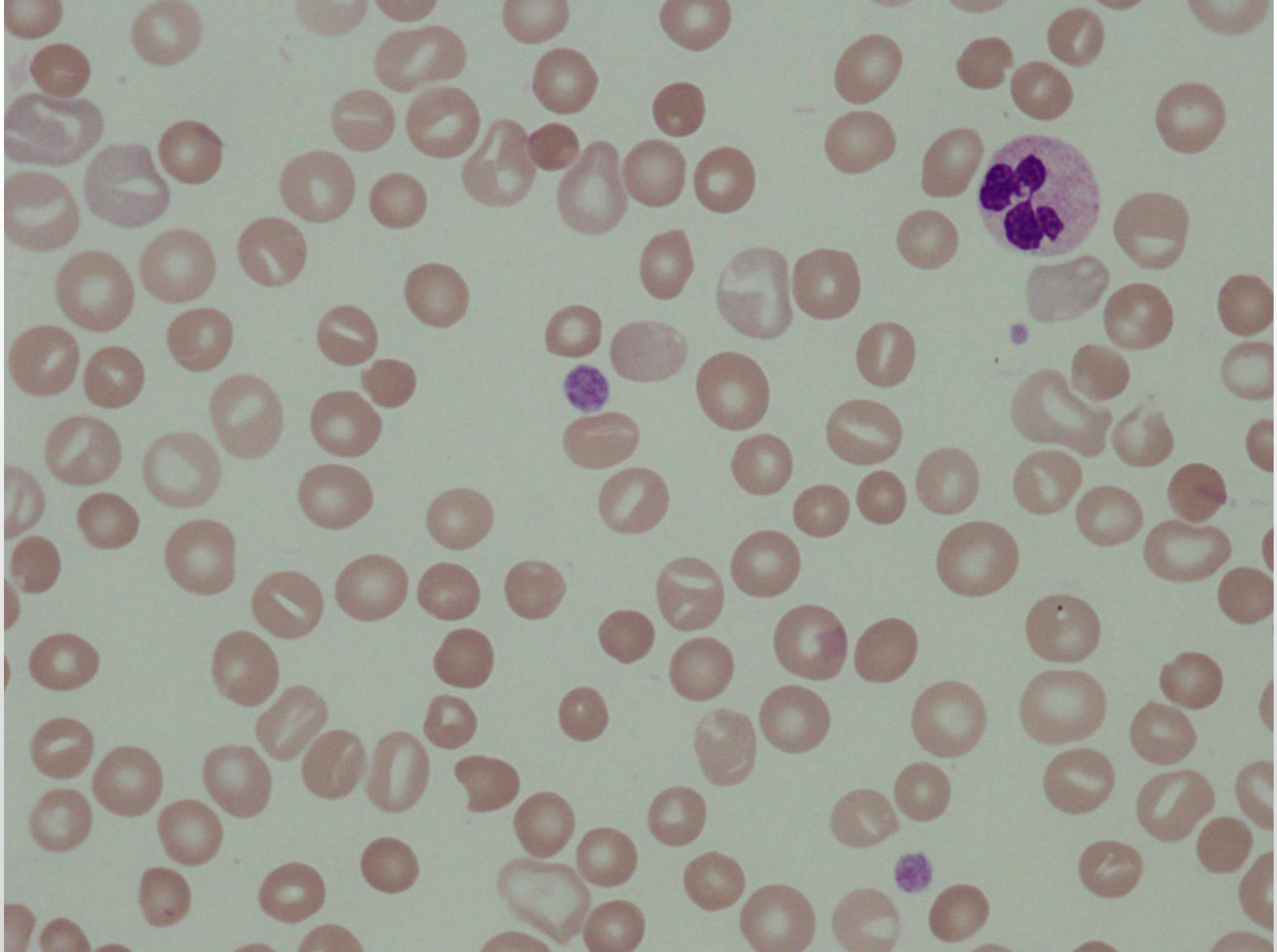
WCC 4.55 x10^9/L

PLT ?









Morphological features

- Giant, normally granulated platelets
- Anaemia
- Polychromasia
- Frequent stomatocytes
- Rare spherocytes, irregularly contracted cells

Final Diagnosis

Possible Mediterranean stomatocytosis/macrothrombocytopenia

Also known as “phytosterolaemia”

- Autosomal recessive
- Rare, first described in Australia in 1969!!
- Due to absorption of excessive plant–derived cholesterol-like molecules
- Multiple ethnicities
- Short stature, xanthomas, high cholesterol
- Variable bleeding phenotype, mild haemolysis
- Abnormal platelet aggregation studies (abnormal ristocetin-induced PLT aggregation, variable to other agonists)