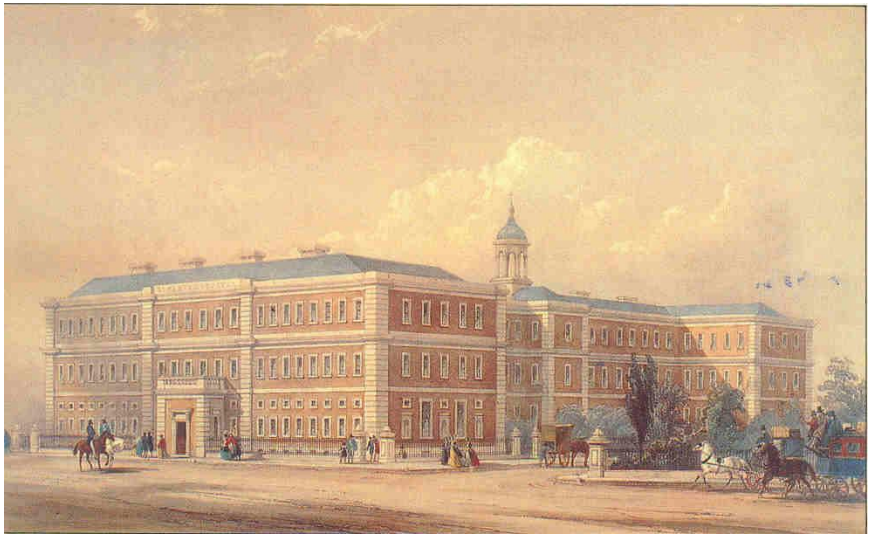
My Favourite Cell – the Spherocyte

Barbara J Bain

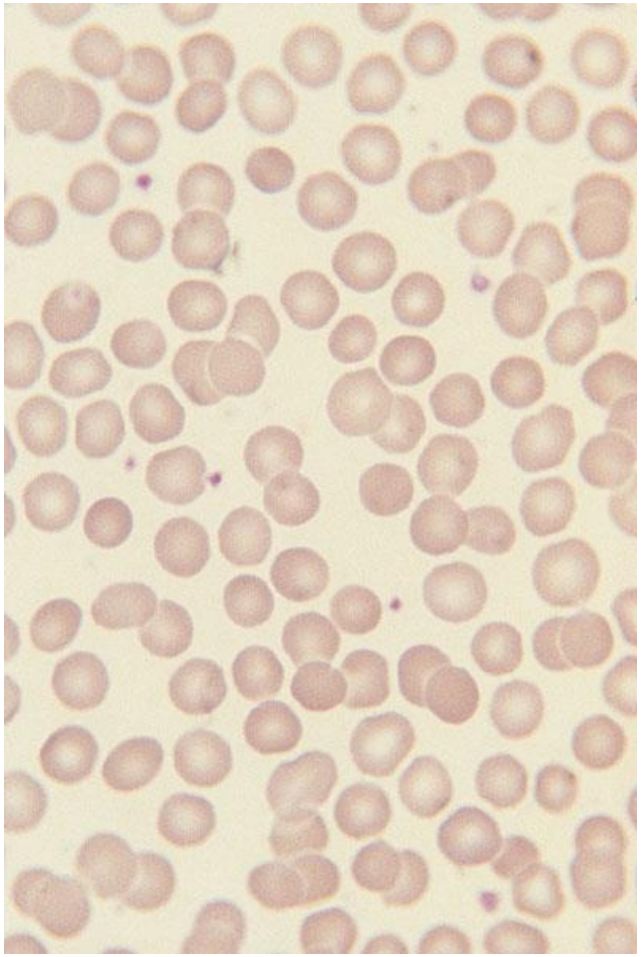


The spherocyte

- Is it a spherocyte? (examine the right part of the film)
- Is it an irregularly contracted cell?
- Is it a microspherocyte/spheroschistocyte?
- If it is a spherocyte, why has it happened?

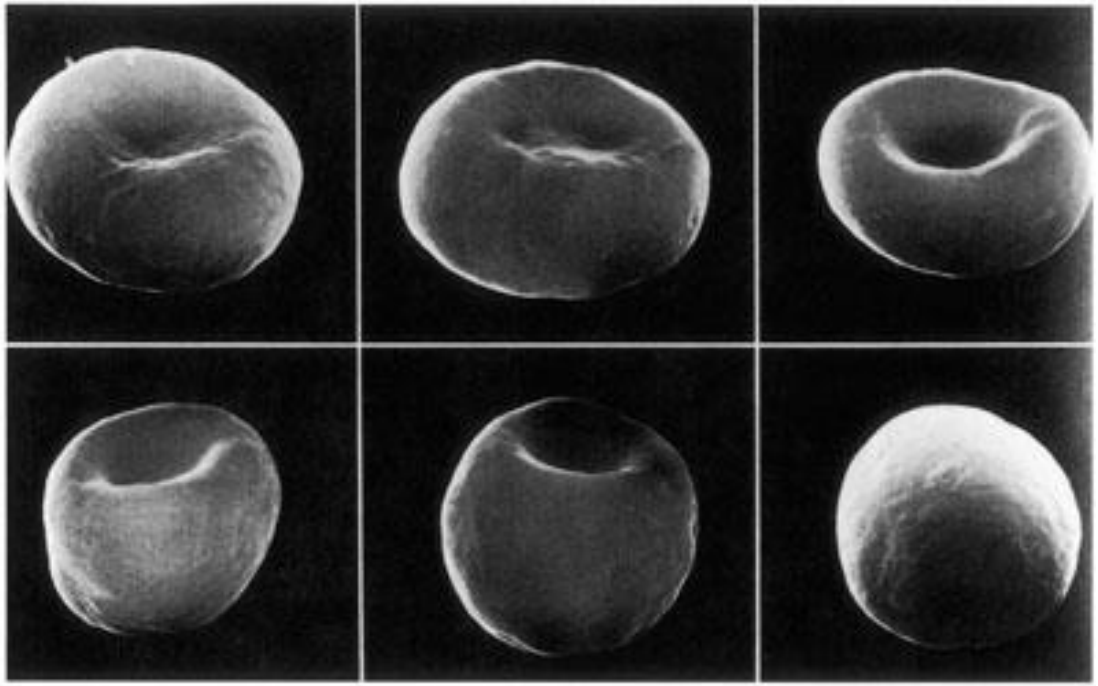
The spherocyte

- It is a spherocyte — hereditary spherocytosis



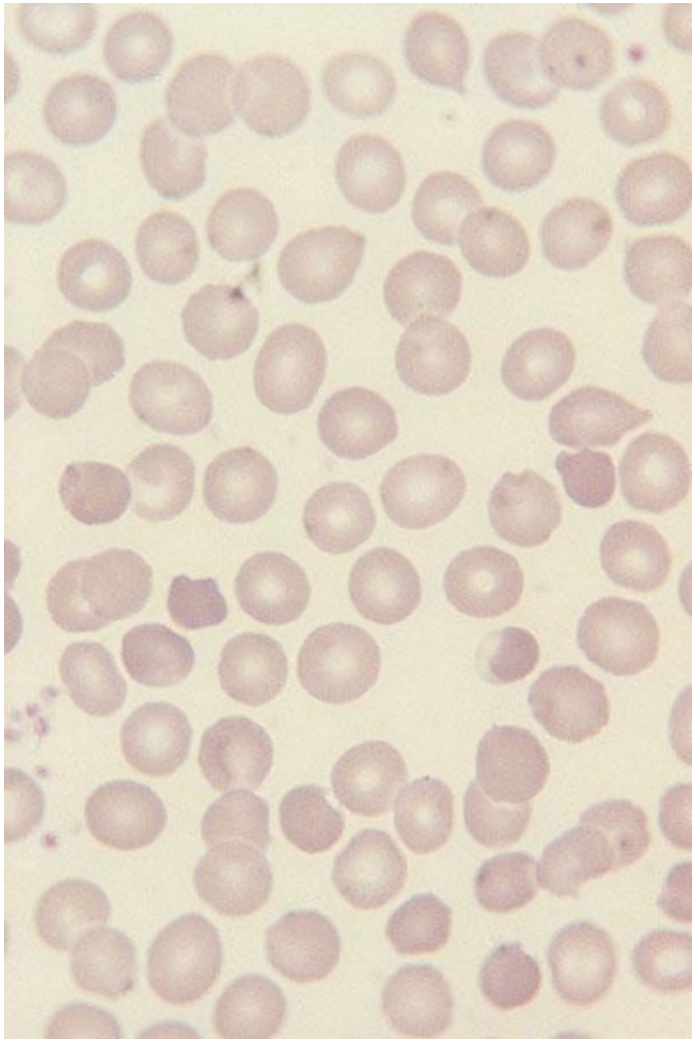
The spherocyte

- Is it a spherocyte?



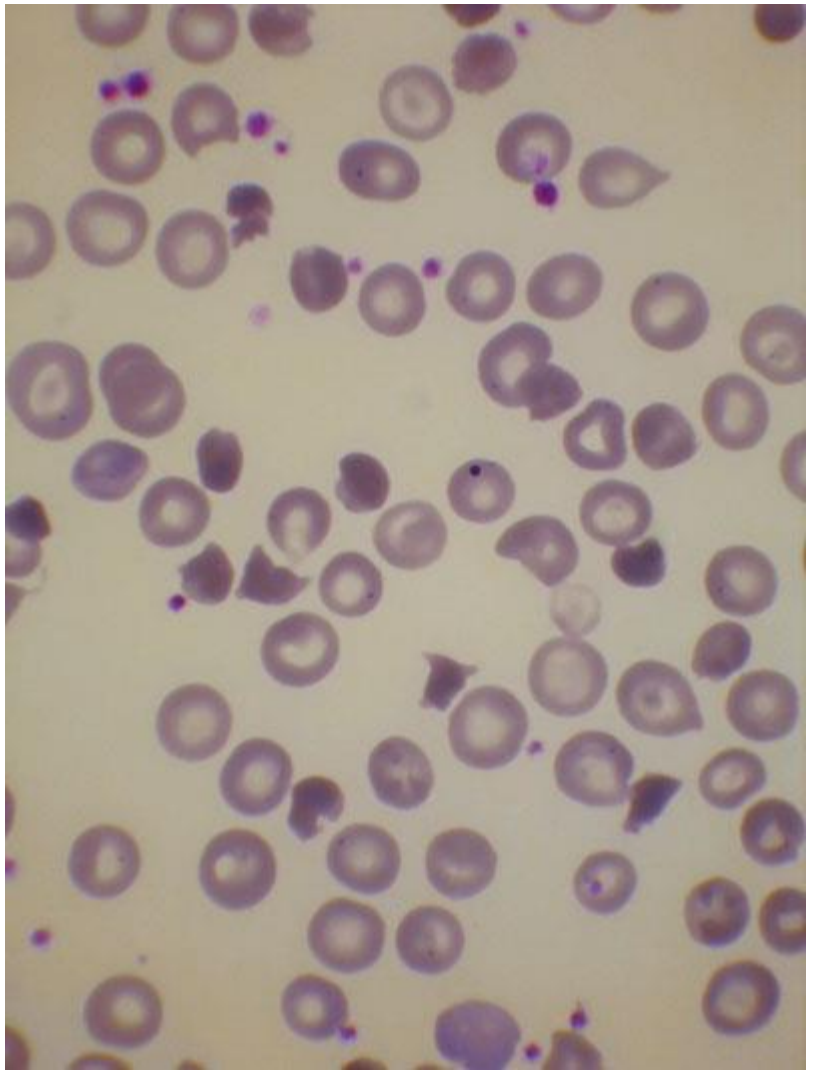
Not a spherocyte

- Not a spherocyte — irregularly contracted cells due to haemoglobin Köln



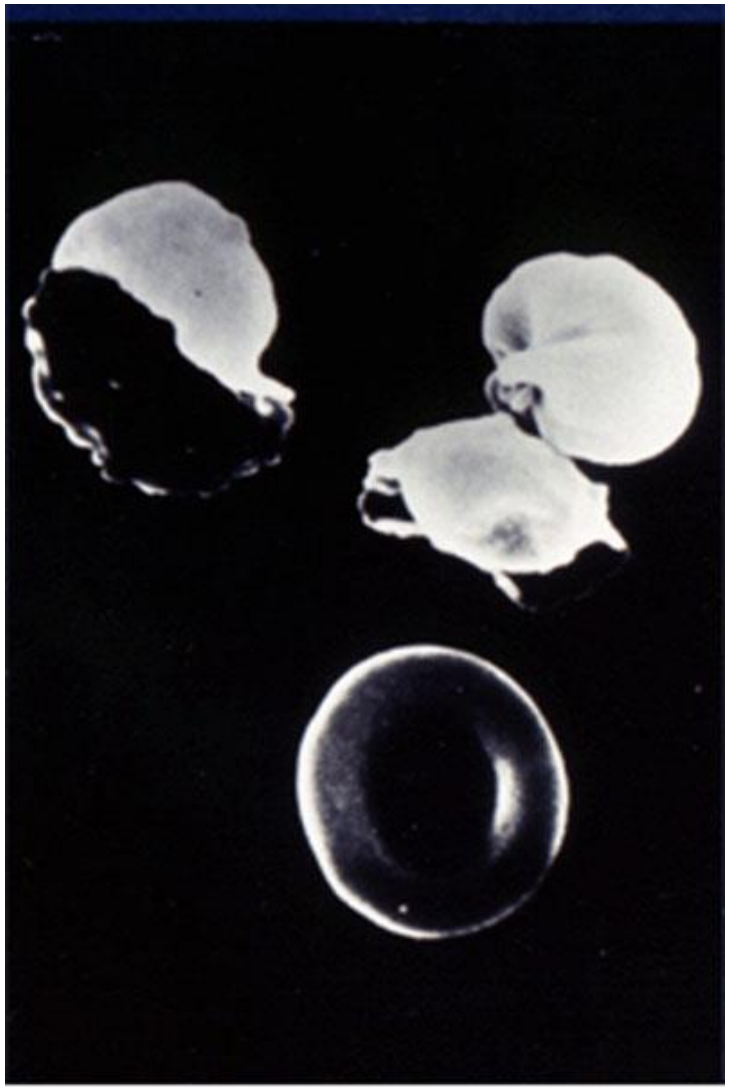
Not a spherocyte

- Not a spherocyte — irregularly contracted cells due to glutathione peroxidase deficiency



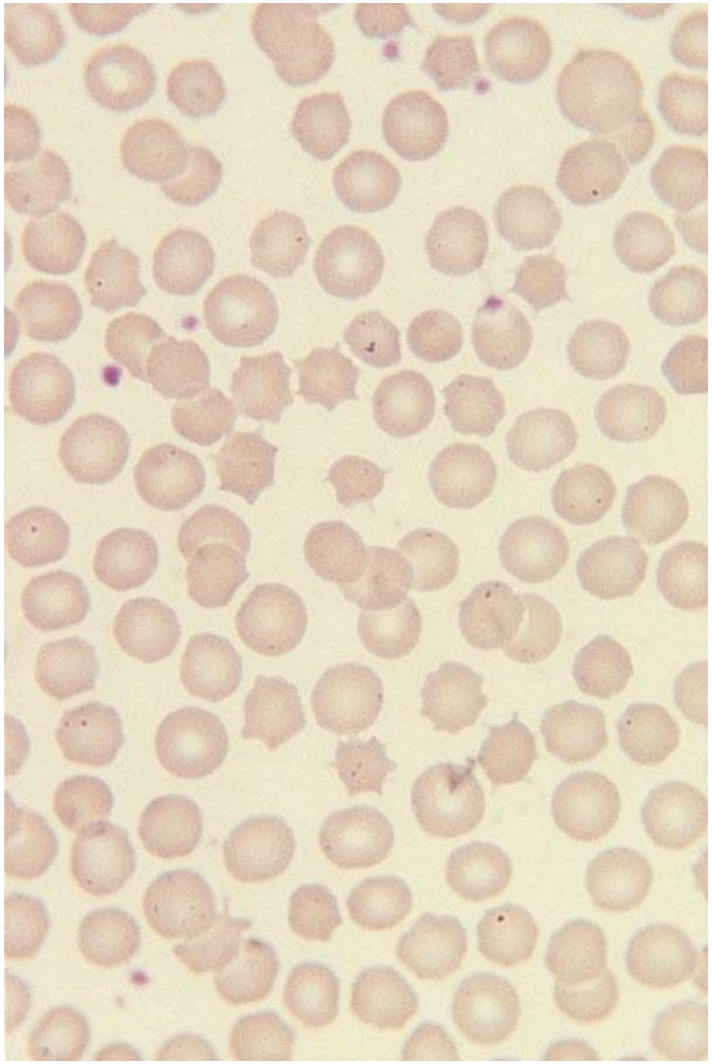
Not a spherocyte

- Irregularly contracted cells due to G6PD deficiency



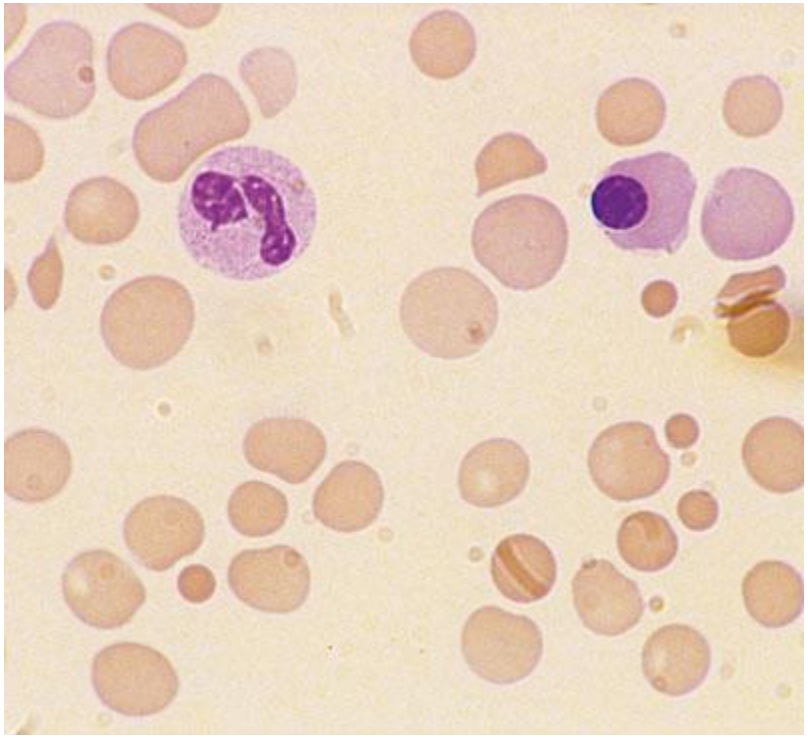
Not exactly a spherocyte

- HS, post-splenectomy — spheroacanthocytes



The spherocyte

- Microangiopathic haemolytic anaemia — microspherocytes

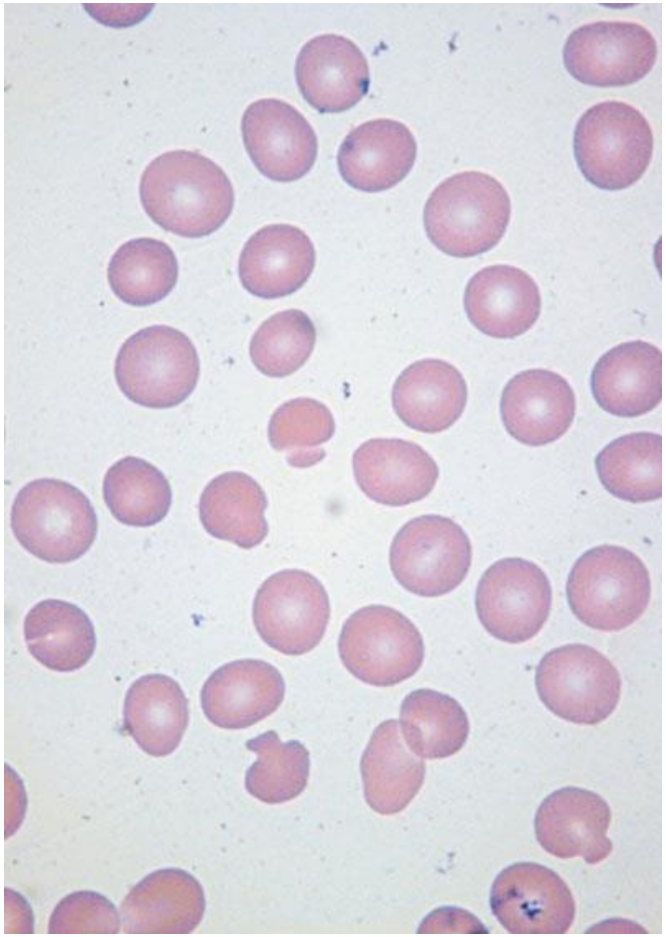


The spherocyte

- If it is a spherocyte, what is the mechanism?
  - Intrinsically abnormal red cell membrane
  - Damage to membrane by an antibody
  - Damage to membrane by a toxin or heat

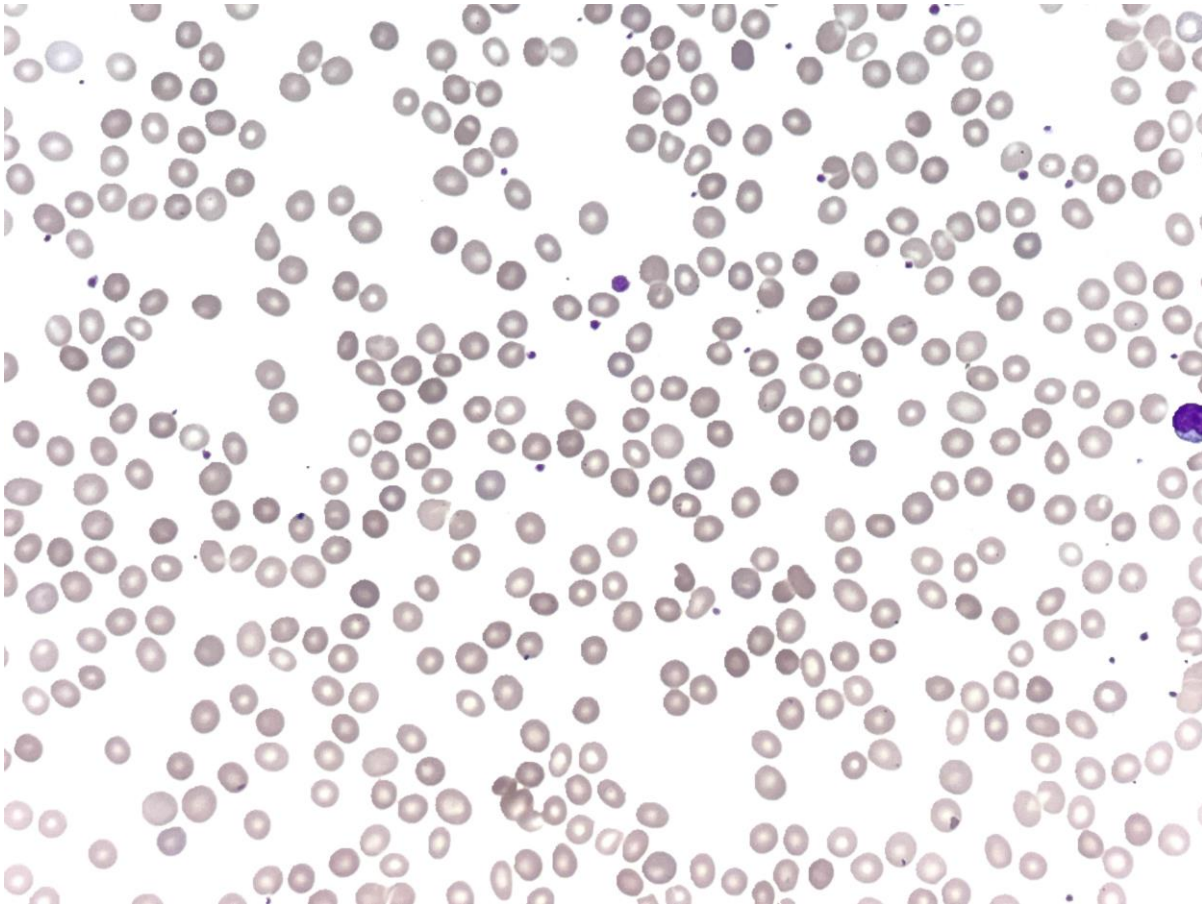
The spherocyte

- Hereditary spherocytosis due to band 3 deficiency



A genetically unusual spherocytosis

- Anaemia
- Spherocytes
- Occasional oval cells and teardrops



A genetically unusual spherocytosis

- The most common forms of hereditary spherocytosis are autosomal dominant with mutations in
  - *ANK1* 50% of cases
  - *SPTB* 30%
  - *SLC4A1* 15-20%

A genetically unusual spherocytosis

- This case is unusual as it is autosomal recessive and due to a mutation in *SPTA1*
- This causes spherocytosis only in homozygosity, compound heterozygosity or when coinherited with a low expression allele

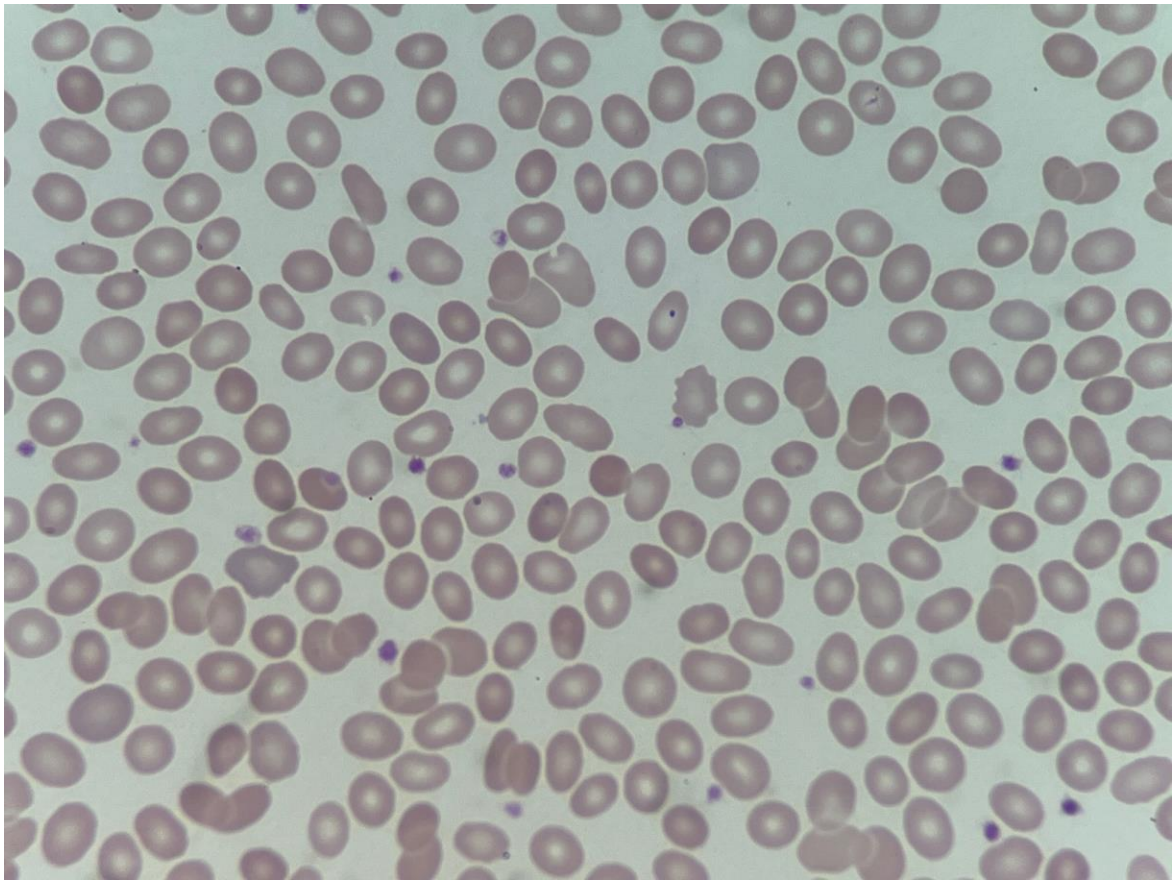
A genetically unusual spherocytosis

- In this patient there was coinheritance of an *SPTA1* mutation and α spectrin^{LELY}
- Of interest the EMA binding was repeatedly normal
- Normal EMA binding does not exclude the diagnosis, which was confirmed by morphology plus genetic analysis

Molina-Arrebola M-A and Bain BJ (2025) Hereditary spherocytosis due to an *SPTA1* nonsense mutation coinherited with α spectrin^{LELY} in *trans. Am J Hematol*, in press

An even more unusul haemolytic anaemia with some spherocytes

- Spherocytes and elliptocytes
- Intermittent anaemia needing transfusion



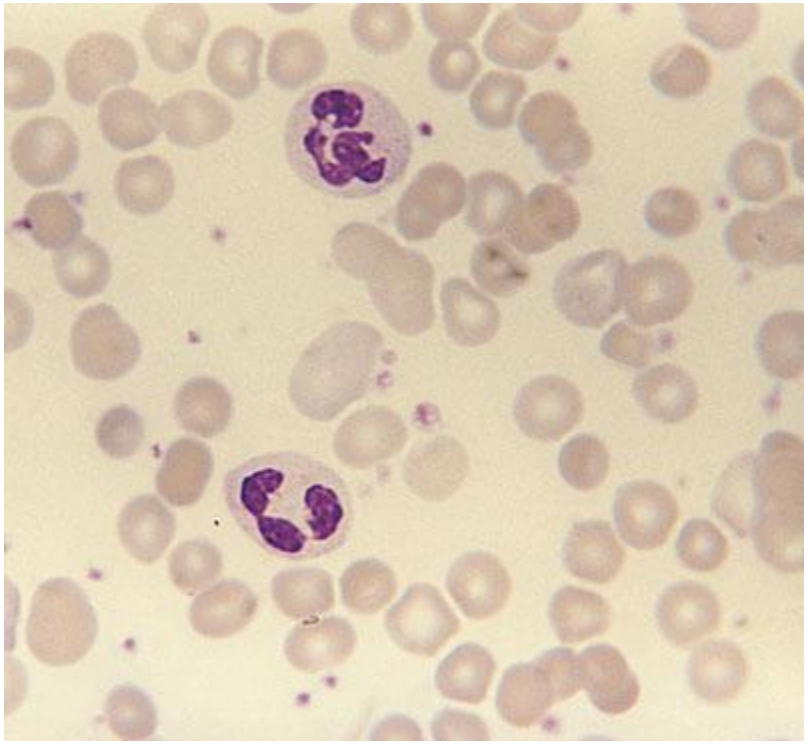
An even more unusul haemolytic anaemia with some spherocytes

- EMA binding abnormal
- This patient had a condition that is sometimes called spherocytic elliptocytosis and sometimes hereditary elliptocytosis
- She was heterozygous for β spectrin Tandil

Molina-Arrebola M-A and Bain BJ (2025) Hereditary elliptocytosis resulting from heterozygosity for β spectrin Tandil. *Am J Hematol*, **9**, 1629–1630.

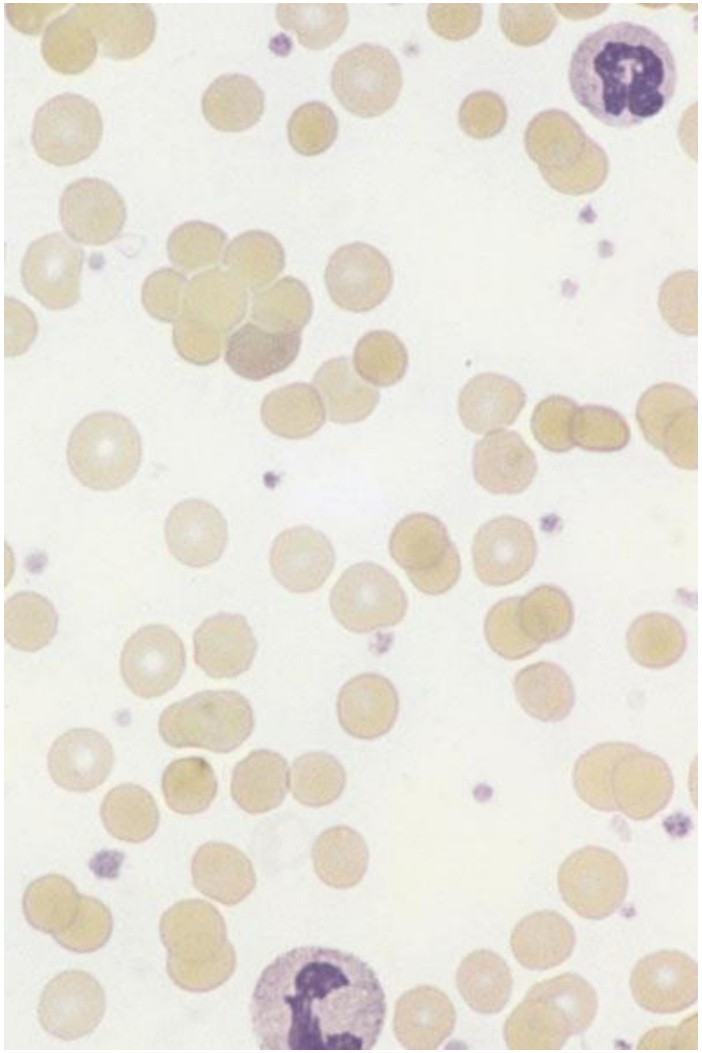
The spherocyte — immune aetiology

- Autoimmune haemolytic anaemia



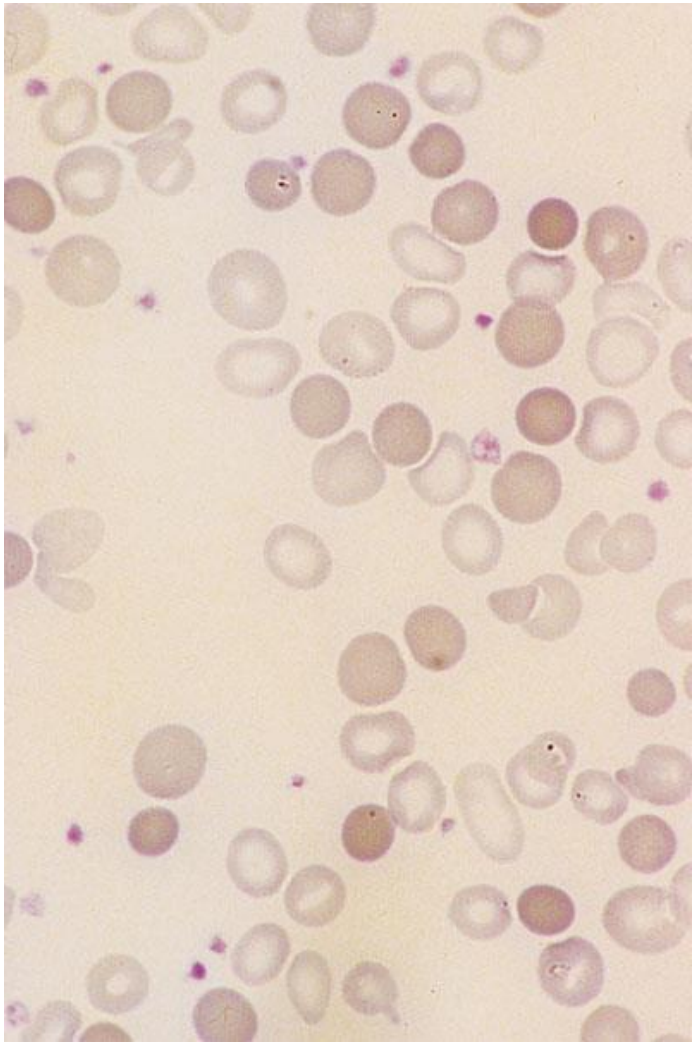
The spherocyte — immune aetiology

- Paroxysmal cold haemoglobinuria



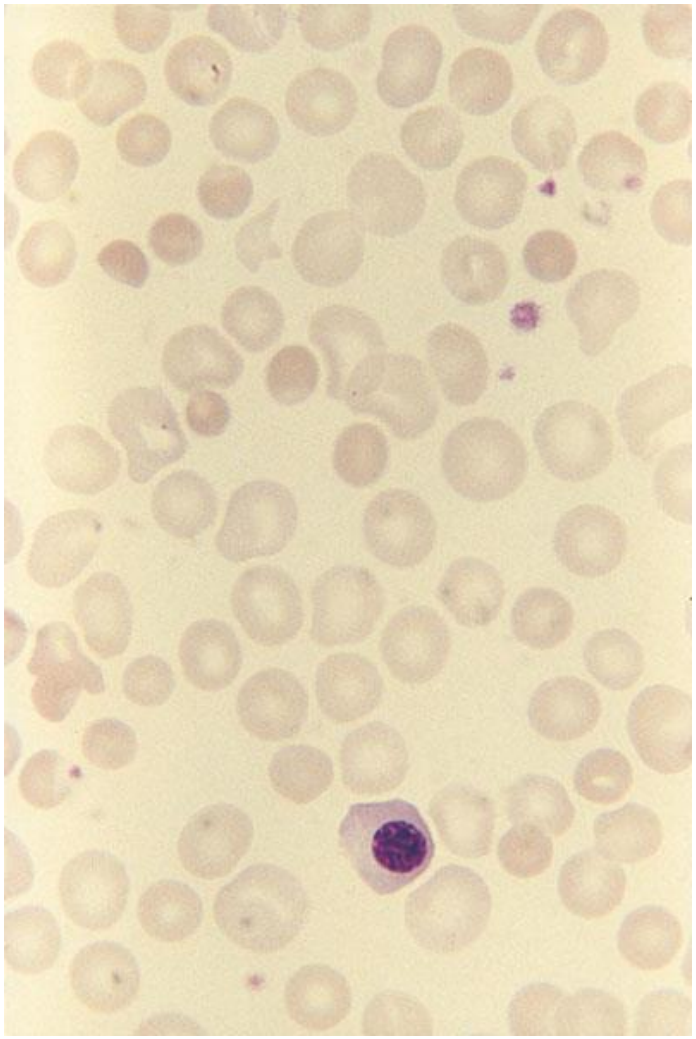
The spherocyte — immune aetiology

- Transfusion of D-positive cells into a D-negative patient



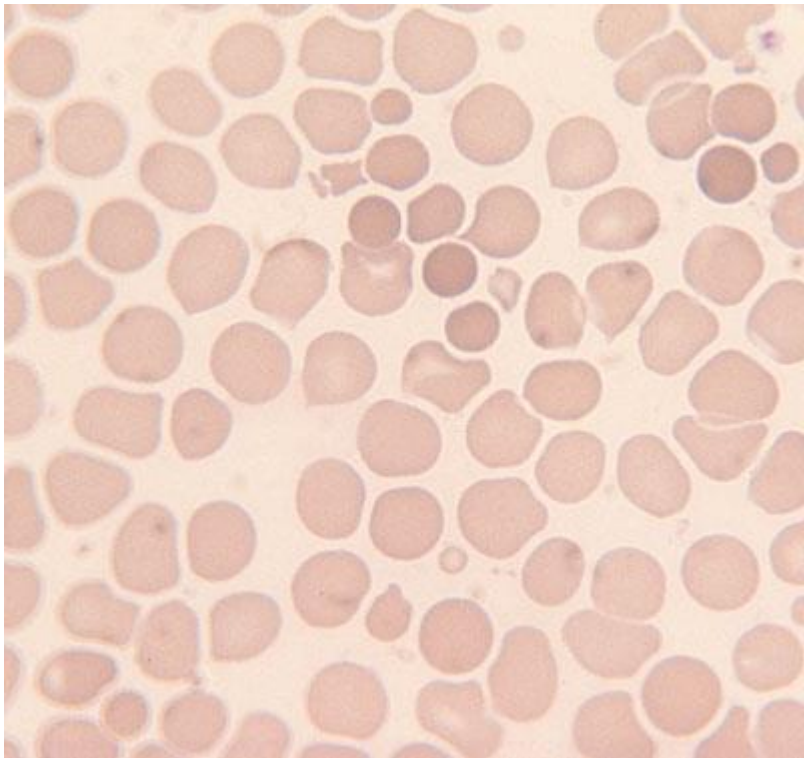
The spherocyte — immune aetiology

- ABO haemolytic disease of the newborn



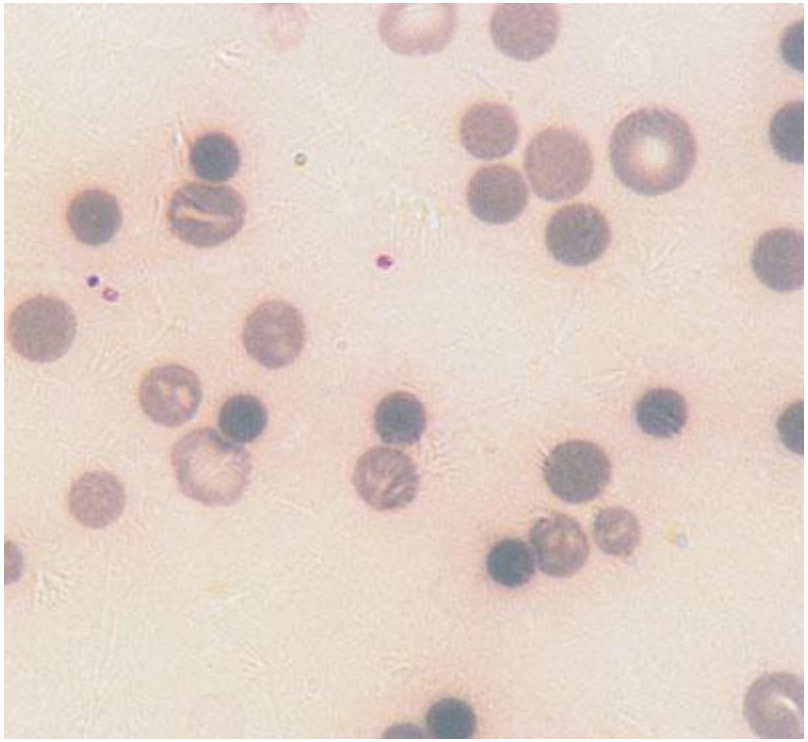
The spherocyte — damage to the membrane

- Microspherocytes in burns



The spherocyte — damage to the membrane

- *Clostridum perfringens*



Conclusions

- Ask yourself two questions
- Is it a spherocyte?
- What is the cause?

The End

Their red cells are elliptical and nucleated