

# Hereditary spherocytosis: a common form of hemolytic anemia, although unexpected when resulting from a “de novo” mutation

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## Introduction

Hereditary spherocytosis (HS) was described in 1871 and the first recorded splenectomy was performed soon after. It is the commonest cause of inherited chronic haemolysis in Northern Europe and North America with a quoted incidence of 1 in 5000 births. However, studies of osmotic fragility in blood donors suggest the existence of extremely mild or subclinical forms, raising the prevalence of HS to 1 in 2000<sup>1</sup>. HS has also been found in other ethnic groups<sup>1</sup>.

Hereditary spherocytosis (HS) is a heterogeneous group of disorders with regard to clinical severity, protein defects and mode of inheritance. It is relatively common in Caucasian populations; most affected individuals have mild or only moderate haemolysis<sup>1</sup>.

<sup>1</sup>Bolton-Maggs PHB; Stevens RF; Dodd NJ; Lamont G; Tittensor P; King MJ. Guidelines for the diagnosis and management of hereditary spherocytosis. British Journal of Haematology, 126, 455-474.

## Clinical Case

A **29 years old man**, with no significant past medical history, was referred to an outpatient Internal Medicine's clinic for:

- **Anemia** (Hb 11,3g / dL, normocytic);
- **Splenomegaly** (17,8x16,5x5,2cm) documented in an abdominal ultrasound.

### He referred:

- **Fatigue**;
- **Asthenia** in the previous year and half;
- Progressively **less tolerance to efforts**.

### He denied:

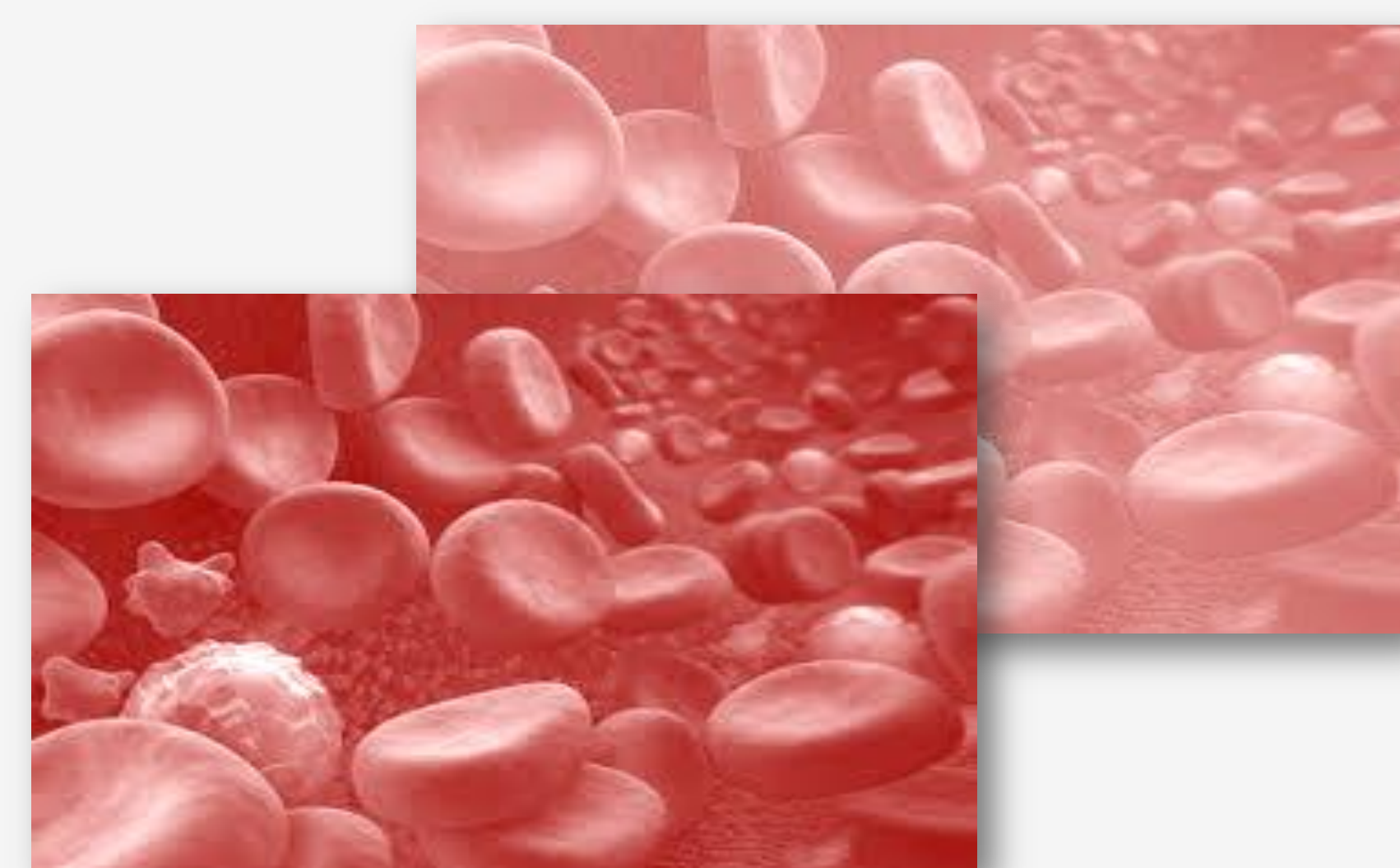
- Blood loss,
- Itching,
- Night sweats,
- Change in color or consistency of stool,
- Change in urine color or jaundice perception
- Family history of anemia

### Physically:

- pale,
- no cyanosis,
- no palpable lymphadenopathy in different ganglion chains ,
- palpable mass in the upper left quadrant compatible with an increased in spleen size.

### Laboratory investigation:

- CBC confirmed normocytic anemia (Hb 12.3 g / dL)
- - Hematocrit (30.2%)
- - White Blood cells (3,9x10x10<sup>6</sup> / uL)
- Haptoglobin <8 mg / dL,
- Reticulocyte count > 3%



### Peripheral blood smear

➔ Red blood cells anisocytosis, poikilocytosis, with a predominance of spherocytes, "bite cells", slight polychromatophilic red cells, compatible with hemolytic anemia for intrinsic red blood cells pathology

### Osmotic fragility (at room temperature)

➔ Increased after 24h

### Genetic study

➔ Mutation for ANK1 gene

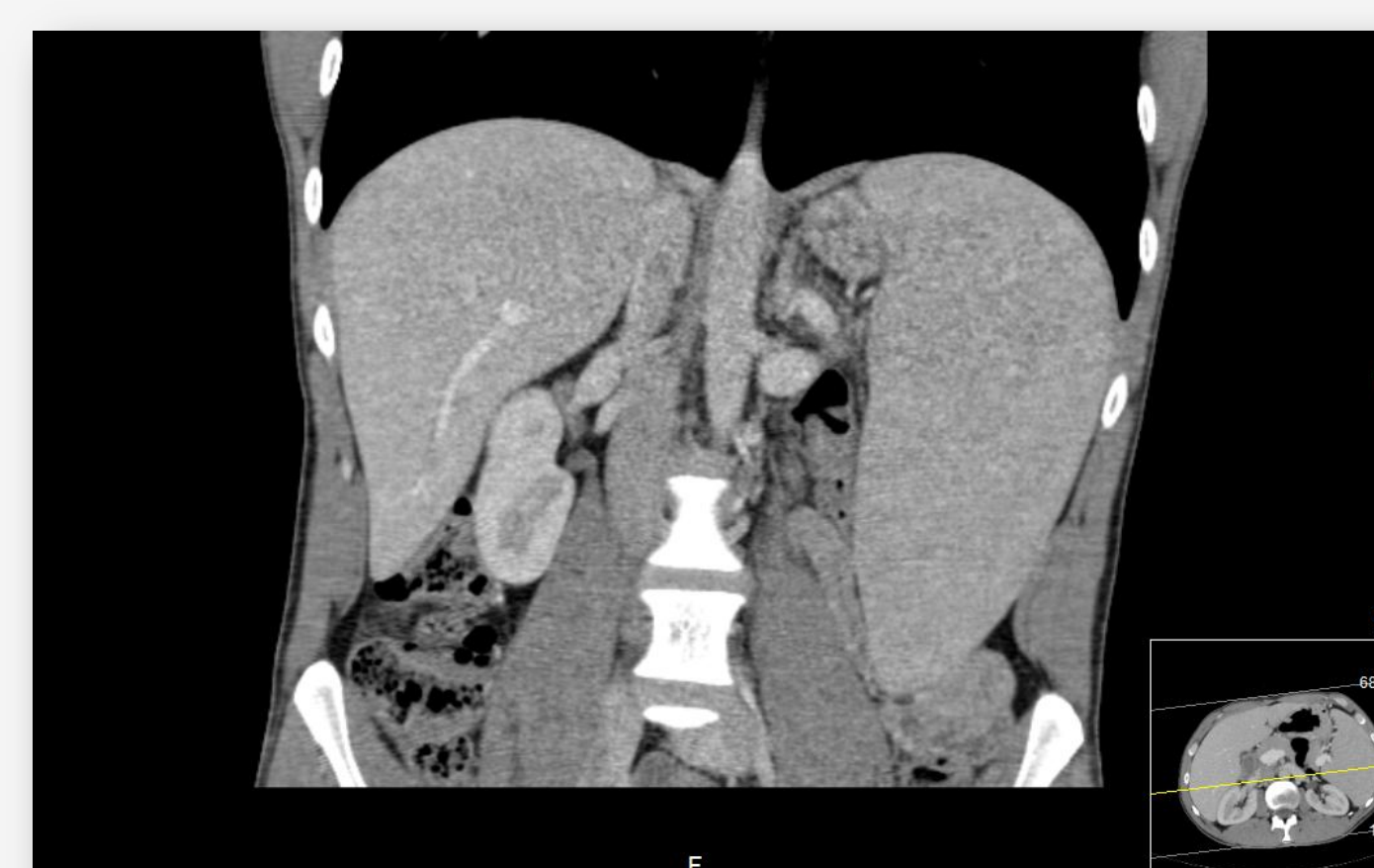
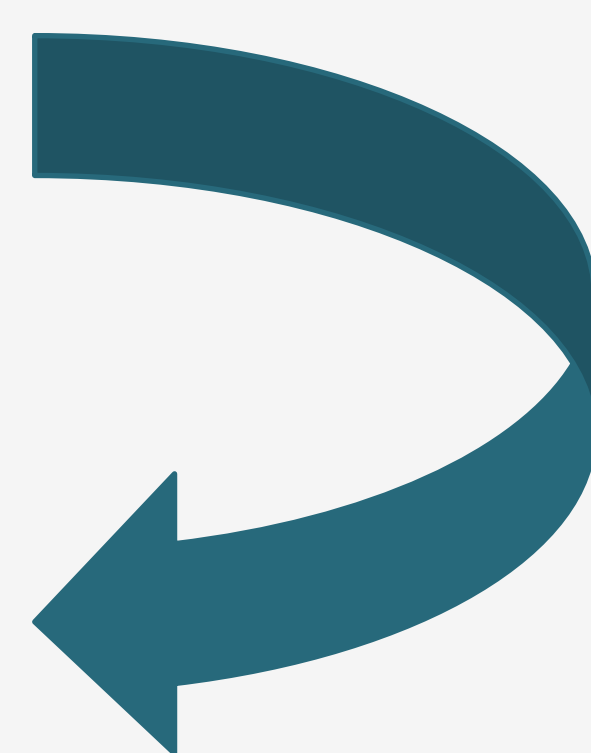
### Upper abdomen computed tomography

➔ Marked splenomegaly spleen with about 19,4cm of craniocaudal diameter and can be a hereditary spherocytosis picture

## Hemolytic anemia

Unequivocally

Hereditary Spherocytosis



## Conclusion

Hereditary spherocytosis is relatively common, with an estimated frequency of 1 in 5000 individuals. Today, we know that is genetically heterogeneous, ranging from mild forms manifesting only in adulthood to severe forms that affect infants catastrophically<sup>2</sup>.

This case illustrates a mild form of disease, which could go unnoticed particularly because of the absence of family history. So, we emphasize the need of a comprehensive and thorough investigation of anemia, for timely intervention, adjusted to the severity of the disease.

<sup>2</sup>Longo DL, Fauci AS, Kasper DL, Hauser SL, Jameson JL, Loscalzo J. Harrison's principles of internal medicine. New York: McGraw Hill, 2011