EUROPEAN HEMATOLOGY ASSOCIATION

EHA-ISHBT Hematology Tutorial

Clinical Case – Session
Transfusion dependent thalassemia

Speaker: V Brousse

Hyderabad,
India
March 1-3,
2024

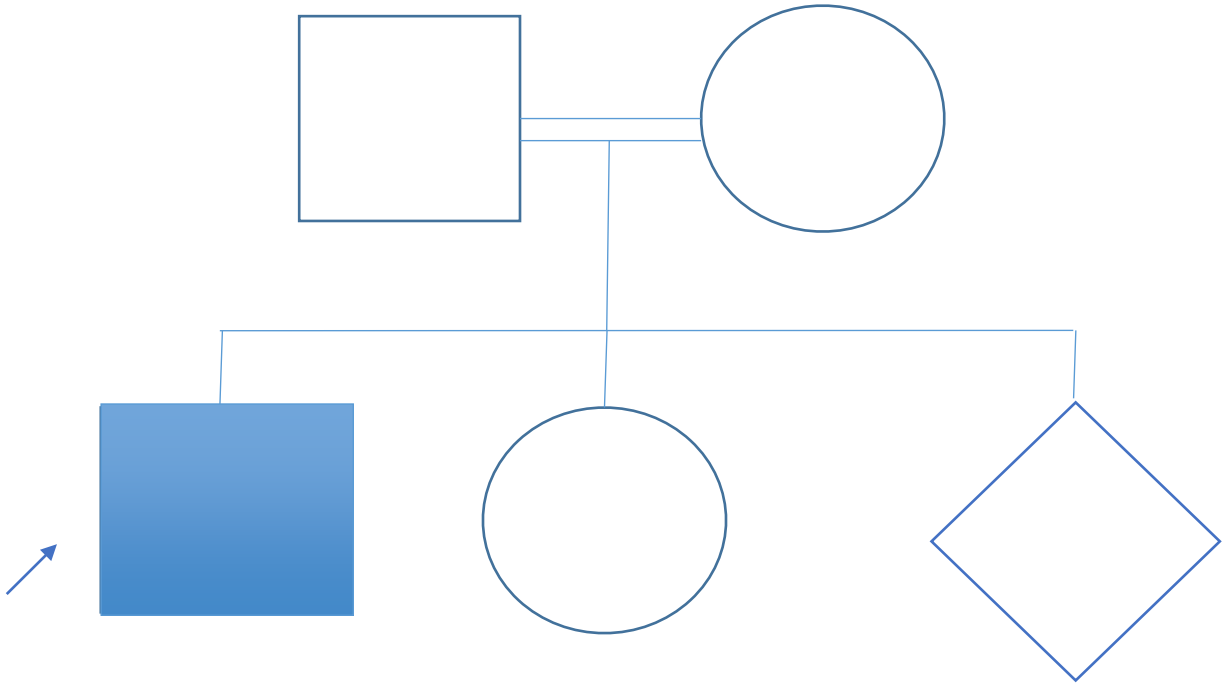
Clinical history

- An 11-year-old boy is referred to you in July 2023 with splenomegaly
- He was diagnosed with β thalassemia following newborn screening

- β globin: homozygous del -1.39 Kb: β°/β°
- α globin: α $-^{3.7}$

Clinical history

- Parents from Mauritania, consanguinous (cousins)



02/06/2016

Clinical history

- First years: apparently uneventful

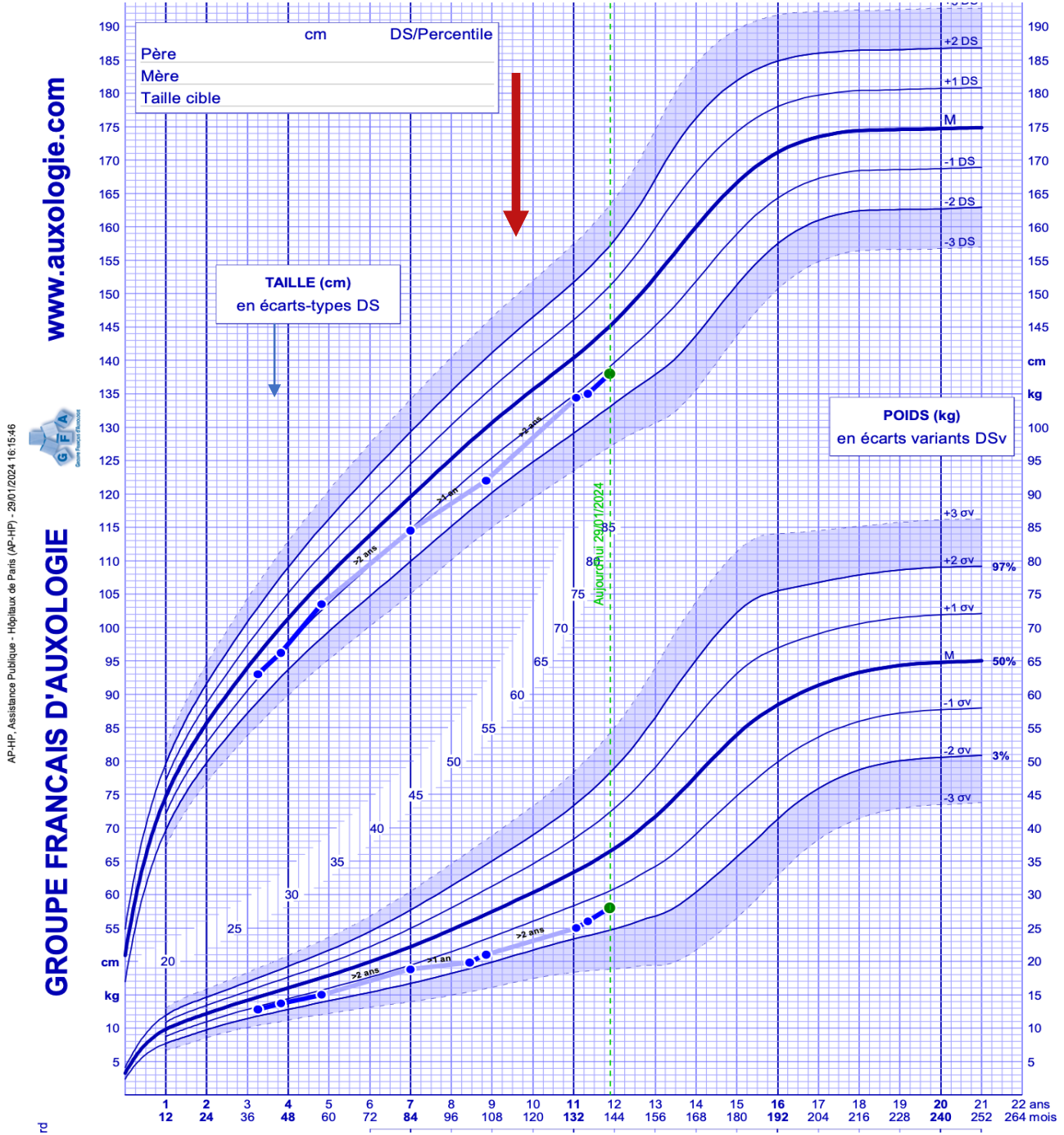
- 2016: 1st transfusion (febrile illness, Hb 53 g/l)
- 2021: 2nd transfusion (febrile illness, Hb 55 g/l)
-
- Splenomegaly noted in July 2016 (2.5 cm)
- January 2017 (Hb 71 g/l)

- **Regular transfusion regimen started in August** 2022 (age 11)

Question

- What important clinical information is missing to assess the severity of β thalassemia in this young patient?

Growth



Transfusion regimen

Clinical history

- July 2023: 1st visit in your clinic, with his mother
-
  - Moderately pale
  - In good general state
  - Splenomegaly 2.5 cm below left costal margin
  - Thalassemic features

Discussion

- What additional important biological information do you need to know to evaluate the clinical situation?

  - Pre- and post-transfusion parameters (FBC)
  - Volume of packed red cells given and interval between transfusions
  - Iron overload parameters
  - Immunization status
  - Viral serologies: hepatitis B, C, human immunodeficiency virus (HIV)

Clinical history

Additional information

- Hb <80 g/l pre-transfusion
- 15 ml/kg packed red cells every 4 weeks
- Ferritin 1250 µg/l

Your advice ?

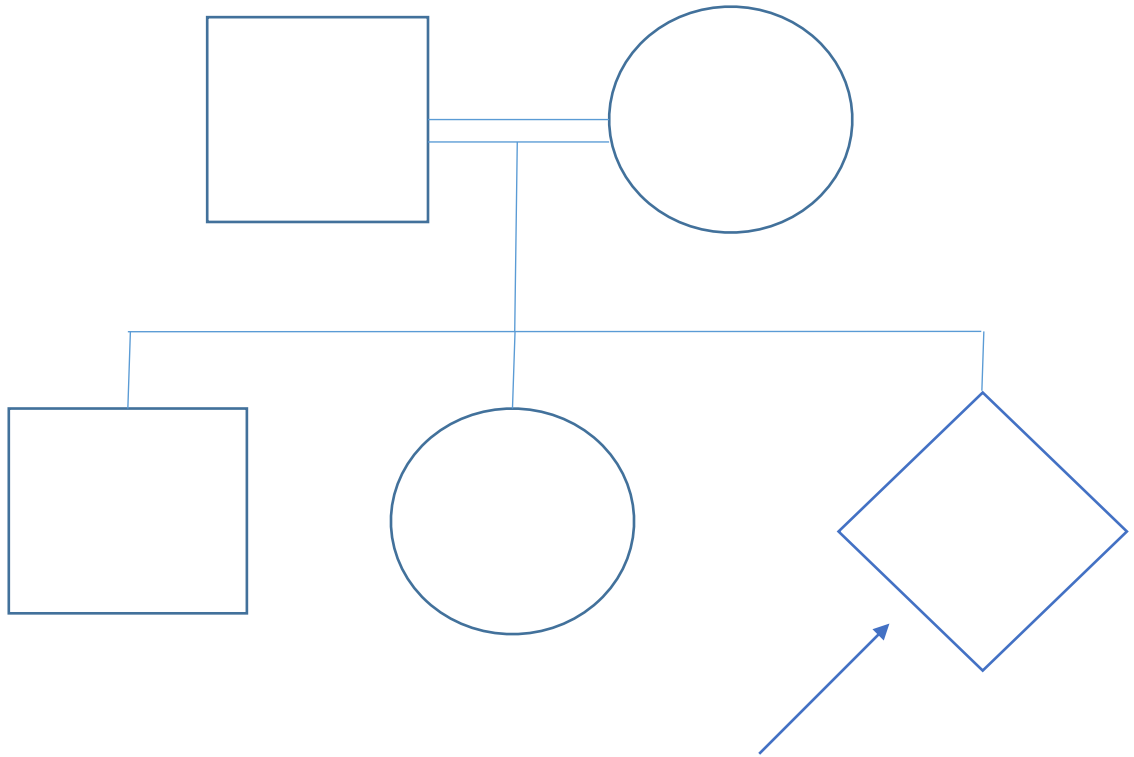
Target >95 g/l pre-transfusion Hb (increase either volume or decrease interval between transfusion)

Perform liver and heart magnetic resonance imaging (MRI)

Start chelation therapy (deferasirox) after pre treatment check-up

Clinical history

- The mother is pregnant
  - Prenatal diagnosis ?
  - Cord blood banking ?
- Sibling HLA typing ?



Clinical history

Second visit in January 2024

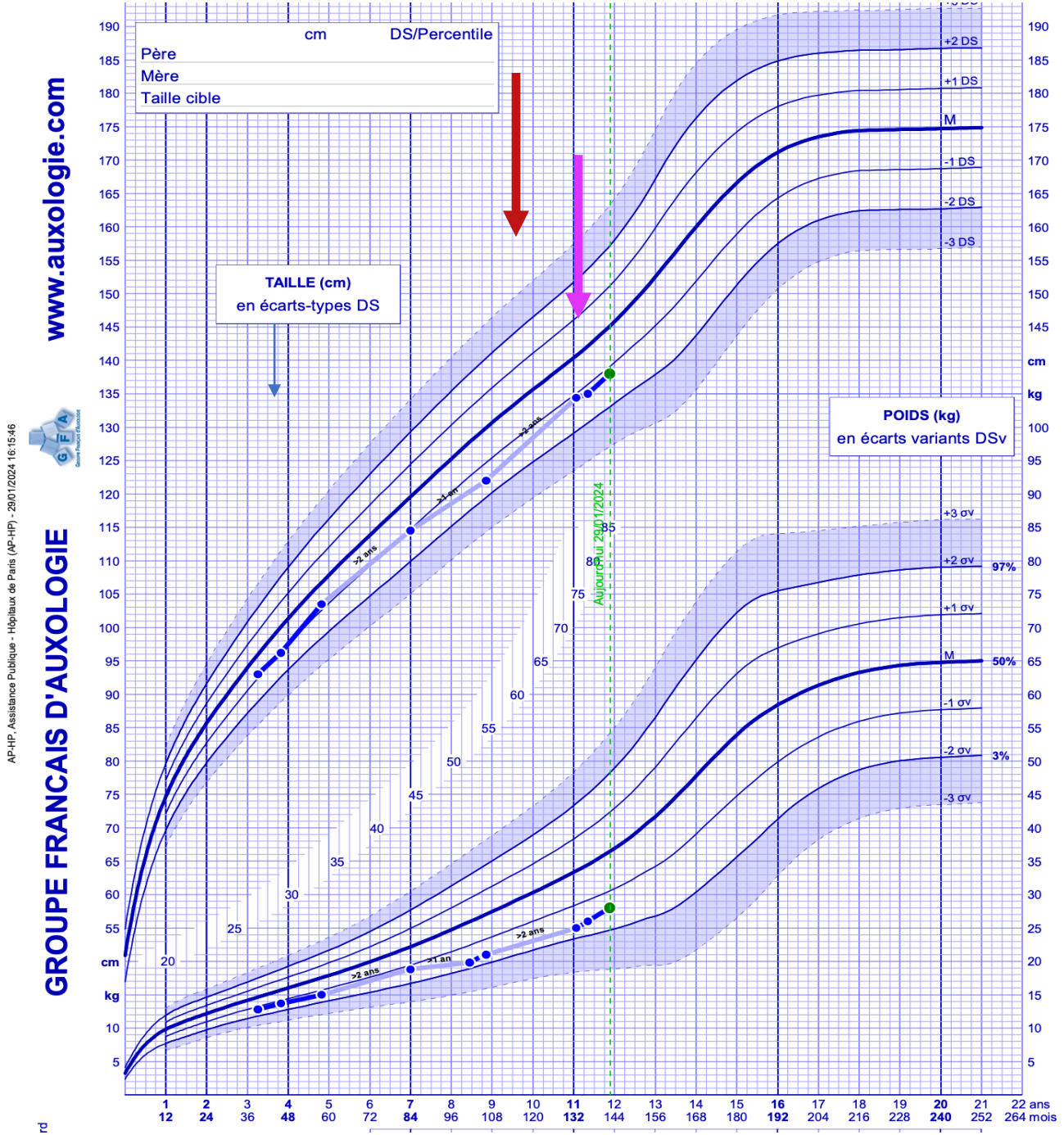
- Hb > 95 g/l (pre-transfusion)
- Transfusion requirements: 180 ml/kg/year
- Regular growth

- Liver MRI: lover iron concentration (LIC) 3 mg/g; heart MRI: T2* 38 ms (November 2023)

- Clinical assessment: **Splenomegaly 5 cm below left costal margin**

Growth

Transfusion regimen



Discussion

Do you recommend splenectomy?

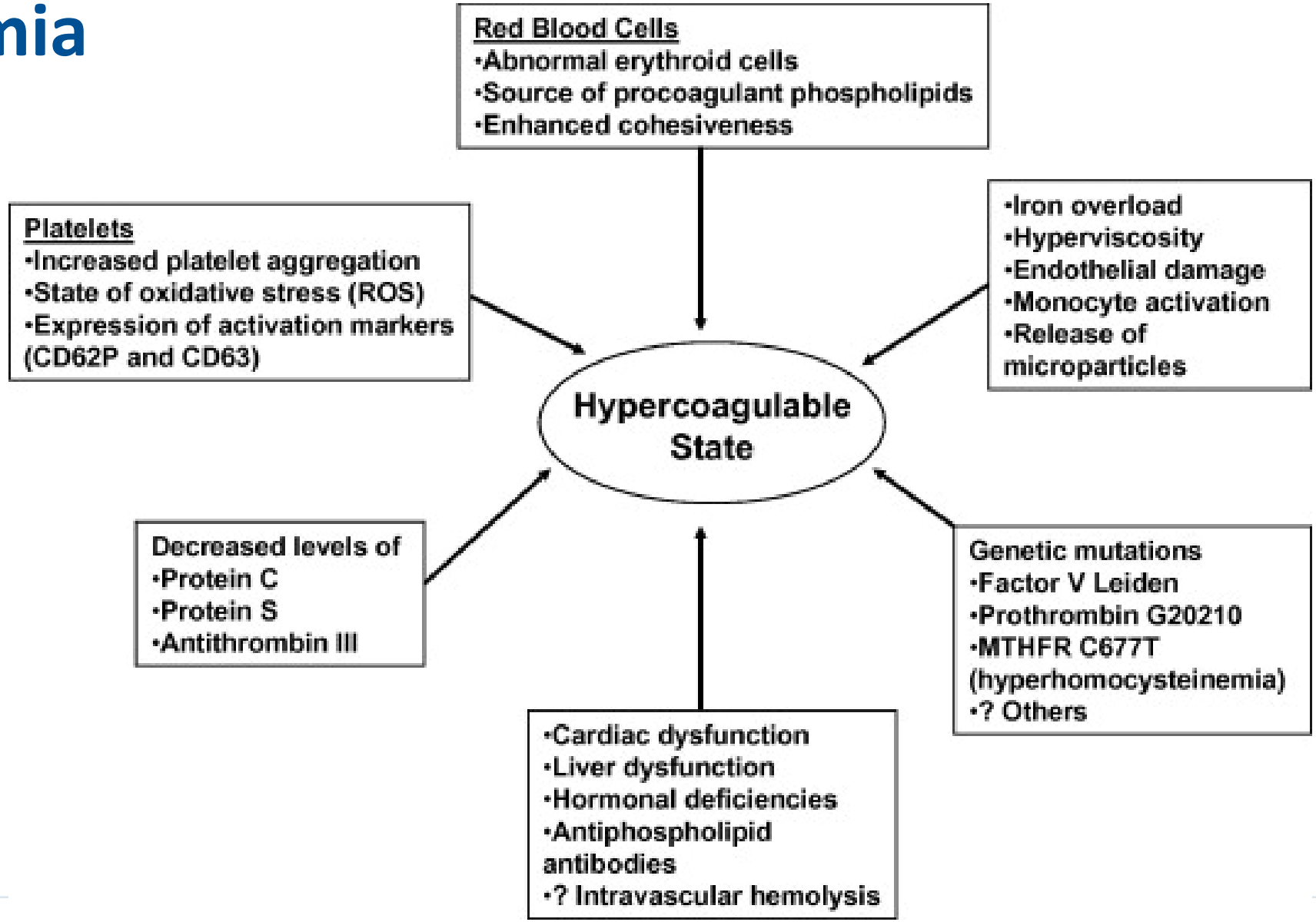
Yes

- Decrease transfusion burden
- Decrease iron overload
- Increase quality of life (QoL)

No

- Moderate transfusion burden
- Increased thromboembolic risk

β Thalassemia



Taher Blood 2005

References

Kattamis A, Kwiatkowski JL, Aydinok Y. Thalassaemia. Lancet Lond. Engl. 2022;399(10343):2310–2324.

Forni GL, Gianesin B, Musallam KM, et al. Overall and complication-free survival in a large cohort of patients with β-thalassemia major followed over 50 years. Am J Hematol. 2022;1-7

Taher AT, Otrock ZK, Uthman I, Cappellini MD. Thalassemia and hypercoagulability. Blood Rev. 2008 Sep;22(5):283-92.

Taher A, Isma'eel H, Mehio G, Bignamini D, Kattamis A, Rachmilewitz EA, Cappellini MD. Prevalence of thromboembolic events among 8,860 patients with thalassaemia major and intermedia in the Mediterranean area and Iran. Thromb Haemost . 2006 Oct;96(4):488-91