

## 9. HEMATOLOGICAL ANOMALIES

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### DEVELOPMENT OF HUMAN HEMATOPOIESIS

Hemoglobin synthesis:

- Embryonic hematopoiesis
- Globin chain “switching”: developmental and environmental regulators
- Embryonic, fetal, neonatal/infant and adult hemoglobins
- Errors in globin chain expression and synthesis
  - $\alpha$ -thalassemias
  - $\beta$ -thalassemias
  - Sickle cell disease
  - Other hemoglobin abnormalities with clinical consequences

*What constitutes a fetal hemoglobinopathy?*

### FETAL ANEMIA

- Etiologies:
  - Hemorrhage
  - Hemolytic anemias
  - Hypoplastic anemias
- Pathophysiology:
  - Intrauterine diagnosis and monitoring
  - Maternal and fetal complications
- Disorders of globin chain synthesis warranting detection in:
  - Prospective parents
  - The fetus

**ALPHA-THALASSEMIA MAJOR (HEMOGLOBIN BARTS DISEASE)**

## Antenatal diagnosis and management

- Multidisciplinary evaluation:
  - Hematological profiles
  - Hemoglobin electrophoretic techniques
  - Molecular diagnosis
  - Placental and fetal ultrasonography
- Associations of Hemoglobin Barts with fetal anomalies
- Rationales for intrauterine therapy
- Intrauterine transfusion (IUT)
- Intrauterine vs. Postnatal hematopoietic stem cell/bone marrow transplantation
  - Immunological barriers
  - Current limits to inducing chimerism
  - Prospects for intrauterine cellular therapies

**CASE DISCUSSION**