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
  - α-thalassemias
  - β-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**