4. GENETICS AND FETAL MEDICINE

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a. MOLECULAR GENETICS

Central dogma
DNA → RNA → Protein
Pre-“genome” era focus
Gene expression, transcription regulation
“Post genome” era
Surprises from the genome
Levels of complexity

b. GENETIC CONTROLS OF DEVELOPMENT

1. Conceptual framework
   • Programming
   • Critical windows
   • Spatio-temporal specificity
   • Genetic Fields

2. Developmental regulations
   • Transcription factors:
   • Specification factors:
   • Transmembrane signaling:
   • Signal Transduction
   • Organizers

3. Example: Cardiac embryogenesis
c. Patterns of Inheritance

1. Autosomal dominant disorders
2. Autosomal recessive disorders
3. X-linked disorders
4. Chromosomal disorders
   - Aneuploidy
   - Deletion syndromes
   - Uniparental disomy
5. Mitochondrial disorders
6. Polygenetic/multifactorial disorders
7. Disruption/deformation/malformation
8. Gene-environment interactions
   - Nutrition, oxygen, toxins

D. Prenatal Diagnosis

1. Preimplantation genetic diagnosis (PGD)
2. Chorionic Villous Sampling
3. Amniocentesis