

4. GENETICS AND FETAL MEDICINE

James F. Padbury, M.D.

a. MOLECULAR GENETICS

Central dogma

DNA → RNA → Protein

Pre-“genome” era focus

Gene expression, transcription regulation

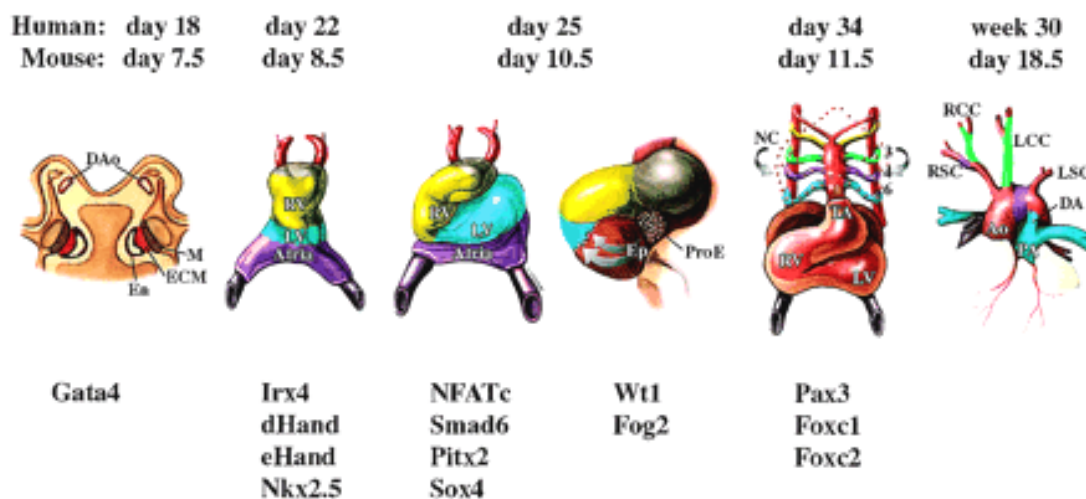
“Post genome” era

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