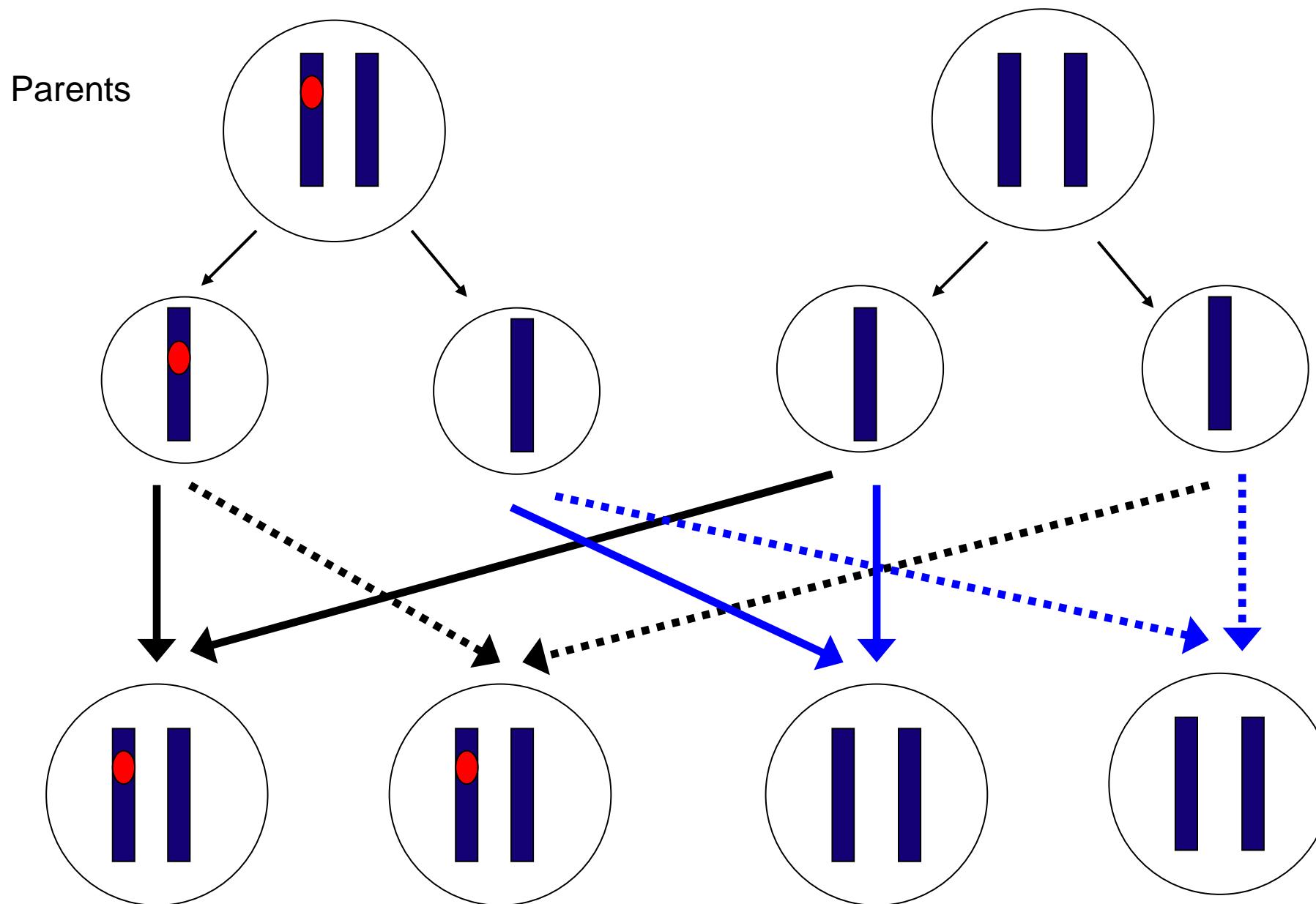
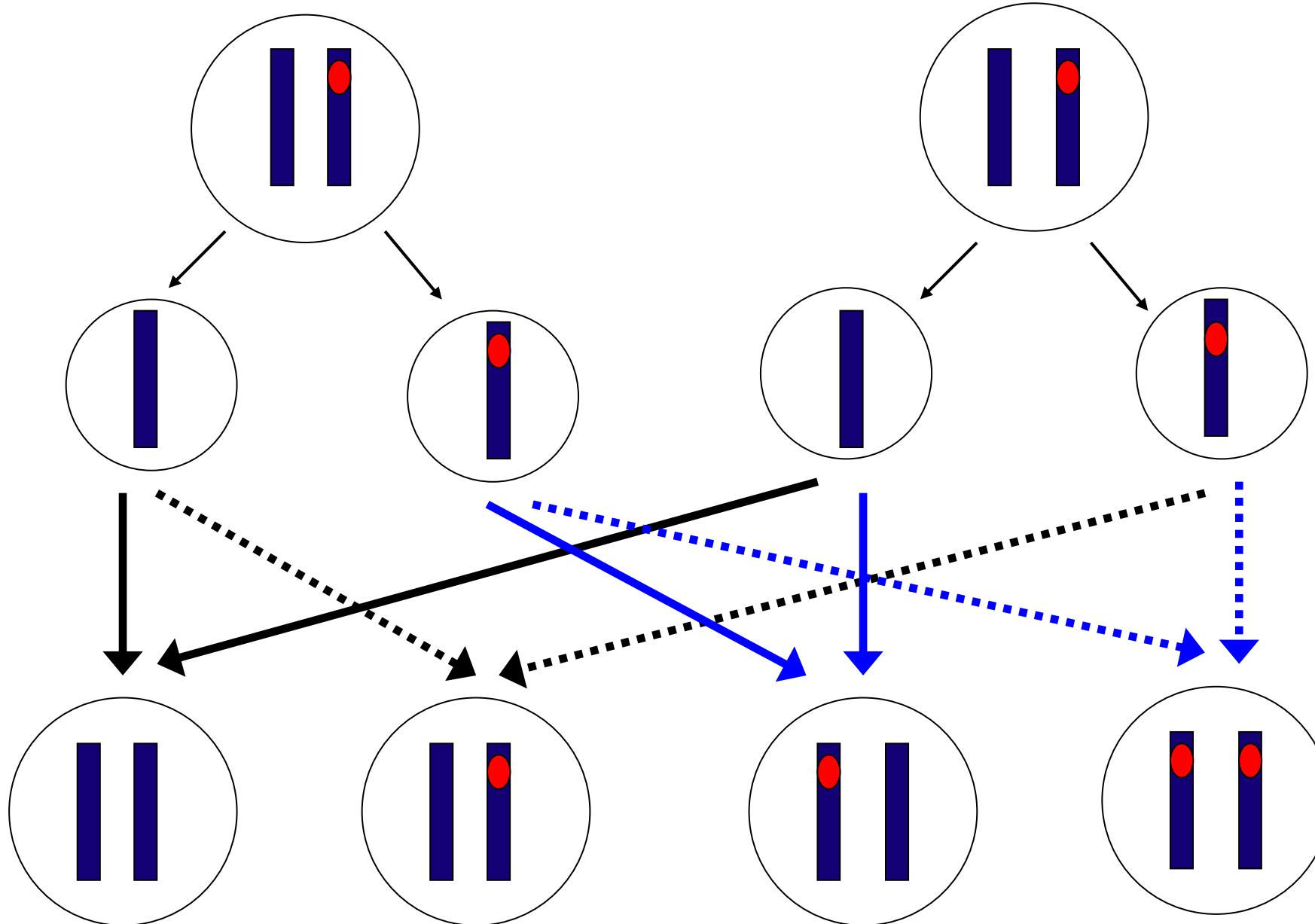


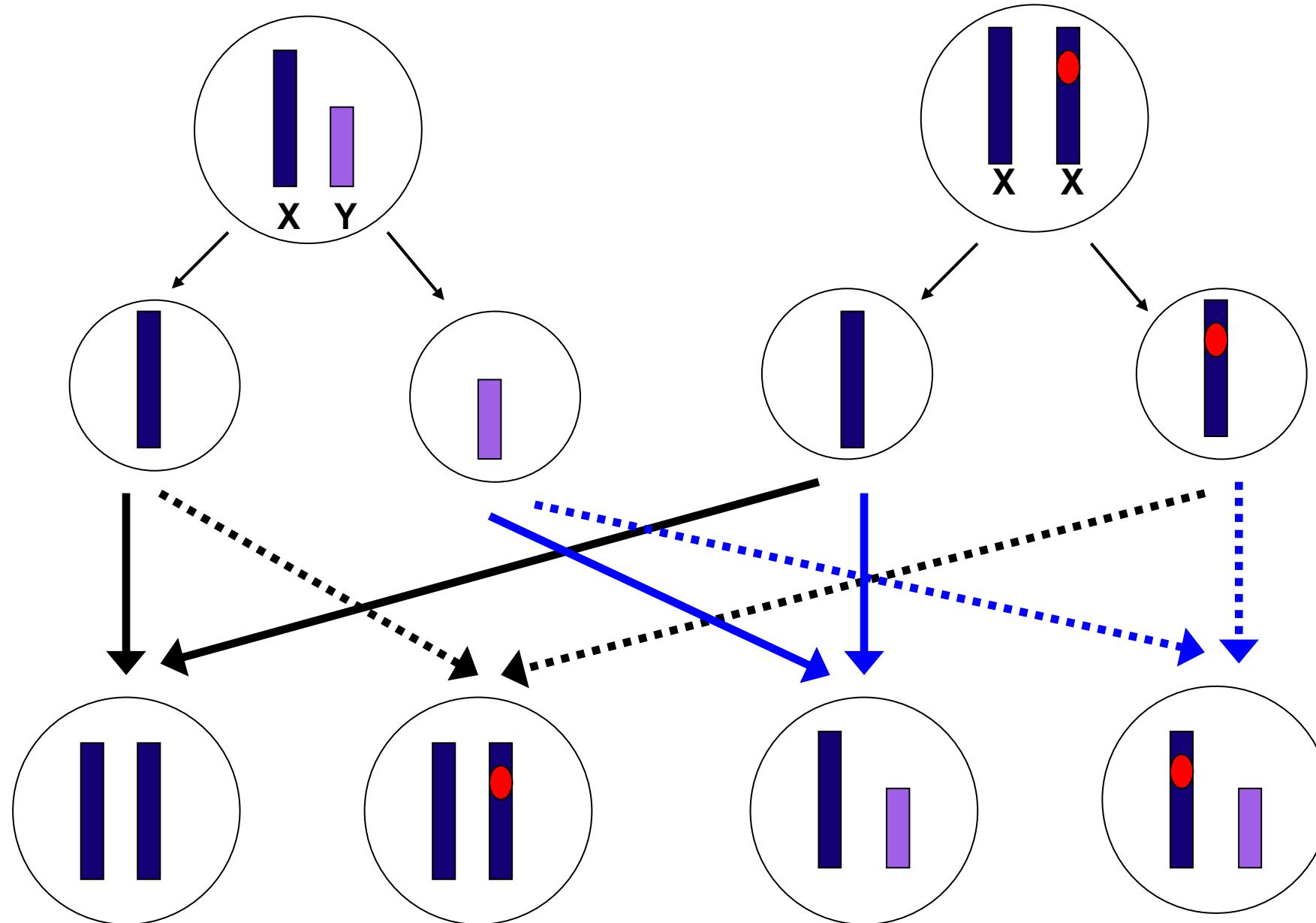
Autosomal Dominant Inheritance

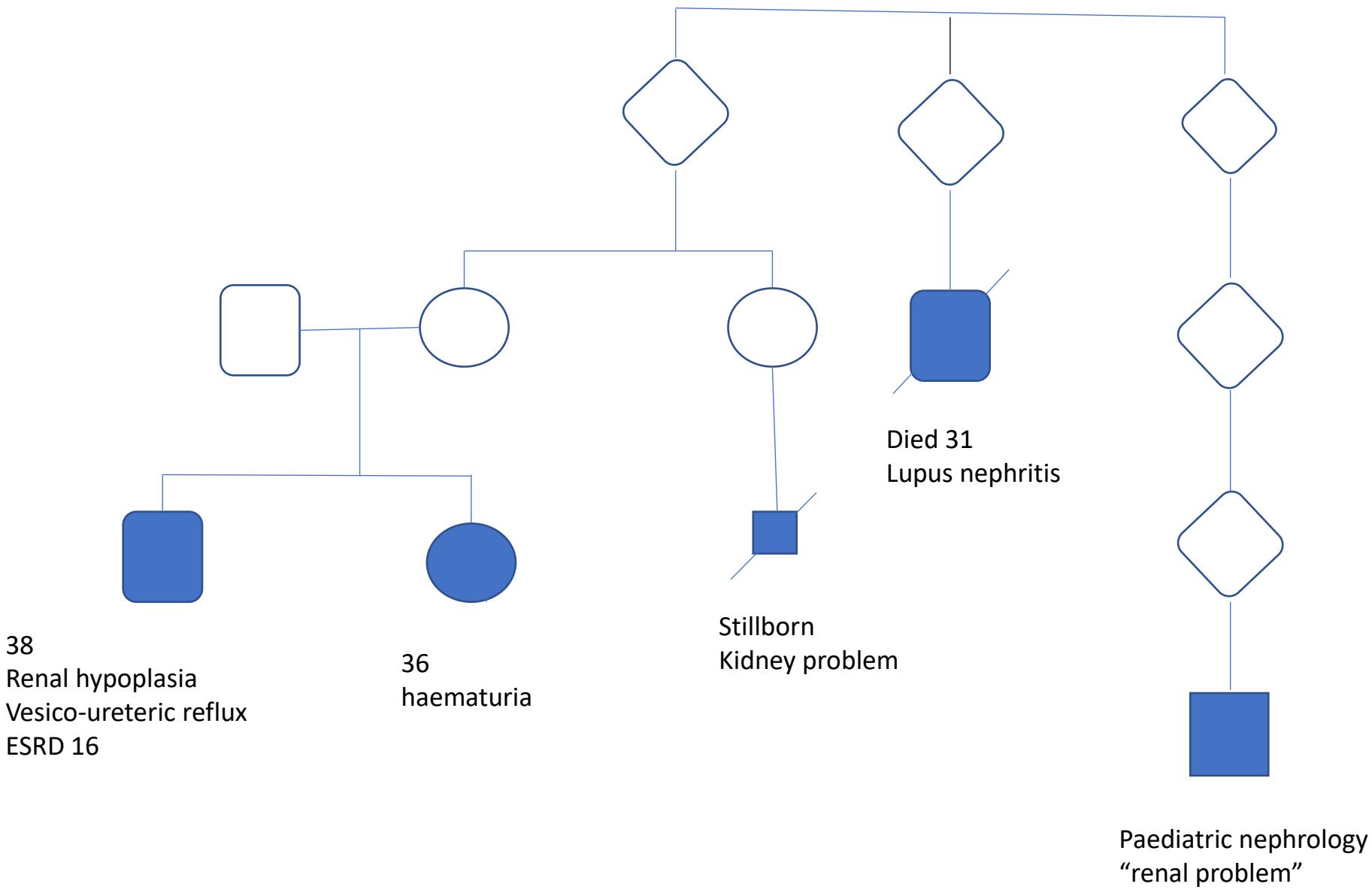


Autosomal Recessive Inheritance



X-Linked Inheritance

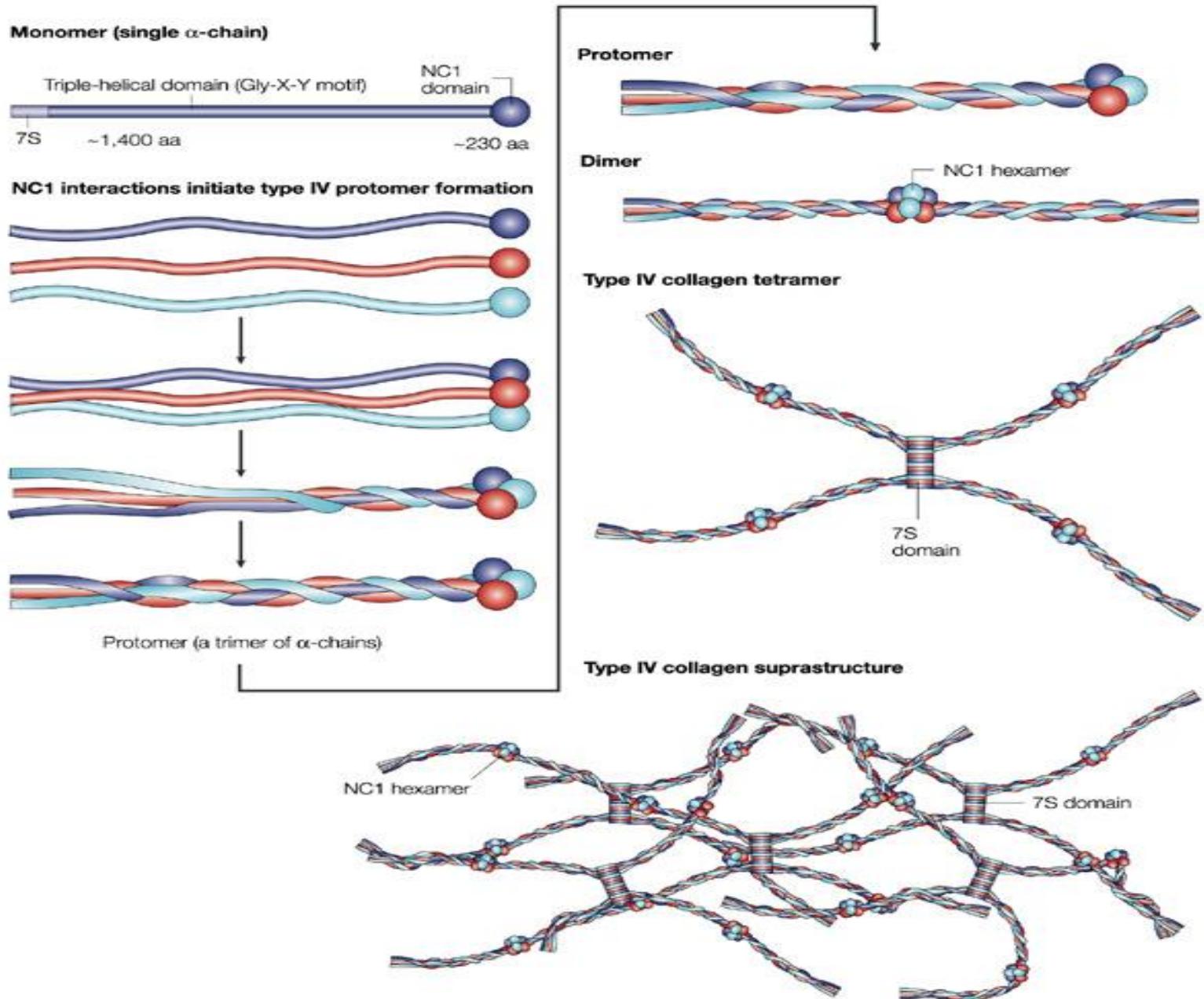




Genetic Testing

- Method
 - Panel test
 - Whole Genome Sequencing
- Type
 - Diagnostic
 - Carrier
 - Predictive

Type 4 Collagen



Alport Disease

- COL4A5
 - X-linked recessive, affected males, carrier females
- Col4A3 & COL4A4
 - Autosomal recessive Alport disease
 - Autosomal Dominant Alport disease
 - AD thin basement membrane nephropathy
 - Genotype/phenotype correlation

The Liability / Threshold Model

