Smith-Lemli-Opitz Syndrome

- May look like “trisomy” disorder
- Main findings:
  - Clenched fists
  - 1-2 toe syndactyly
  - Postaxial polydactyly
  - Cataracts
  - Ambiguous genitalia or sex reversal (i.e., XY female)
  - Hirschsprung’s
  - Mental retardation
- Autosomal recessive; defect in cholesterol synthesis due to mutations in 7-dehydrocholesterol (DHC) reductase
- Diagnosis by showing extremely elevated 7-DHC in plasma
Colostomy due to Hirschsprung’s; 2-3 toe syndactyly, prominent palatal ridges
Ambiguous genitalia, vestigial polydactyly in 46, XY male