

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