Condition: Smith-Lemli-Opitz syndrome

Inheritance:
Autosomal recessive.

Genetic etiology:
Mutation in *DHCR7* gene, which encodes 7-dehydrocholesterol reductase.

Frequency:
1/20,000 – 1/40,000 births.

Clinical features:
Smith-Lemli-Opitz syndrome is a multiple congenital anomaly disorder due to failure in cholesterol synthesis due to deficiency of 7-dehydrocholesterol reductase. Features include growth retardation, microcephaly, severe mental retardation, cleft palate, cardiac defects, polydactyly, syndactyly of the second and third toes, and underdevelopment of male genitalia.

Management:
Supportive care; dietary supplementation with cholesterol.

Genetic counseling:
Based on autosomal recessive inheritance; molecular genetic testing available.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.