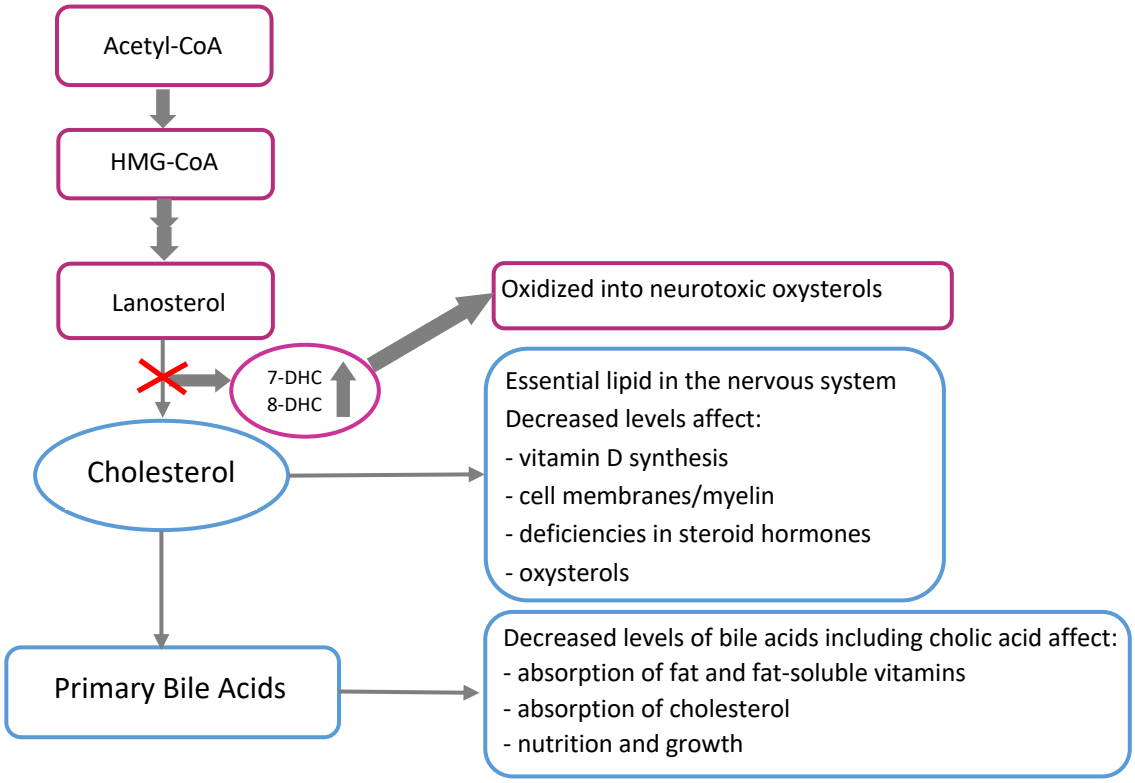
At a Glance
Smith-Lemi-Opitz Syndrome (SLO)



Background[1]

Prevalence: 1 in 40,000 live births

Inheritance: autosomal recessive

Deficient enzyme (gene): 7-dehydrocholesterol reductase (*DHCR7*)

Toxic Metabolites: 7-dehydrocholesterol (7-DHC) and (8-DHC) 8-dehydrocholesterol

Impact: low circulating cholesterol and bile acids

Clinical presentation: prenatal and postnatal growth restrictions, microcephaly, 2-3 syndactyly of the toes, cleft palate, genital anomalies (males), intellectual disabilities, behavioral difficulties, cataracts, abnormal liver function, skin photosensitivity

Primary dietary treatment: cholesterol supplementation

Goal of Treatment: to increase cholesterol levels systemically, limit 7DHC and 8DHC accumulation, and improve physical and developmental outcomes

Medical Therapy

Cholic acid therapy: CHOLBAM 10 – 15 mg/kg by mouth, once daily or in two divided doses, in pediatric patients and adults (mirumpharma.com)

- Utilized to improve dietary cholesterol absorption, liver enzymes, and may positively impact growth
- Pilot study showed improvement in plasma cholesterol and decreased 7-DHC after 2 months[2]

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Nutrition Therapy

Goal: Promote growth and improve plasma cholesterol
Macronutrients: meet age appropriate needs to support growth, based on SLO growth charts[3]
Diet: High cholesterol, high fat diet
High cholesterol foods
1 large egg yolk = 185 mg cholesterol, 4.5 g fat
1 oz heavy cream = 34 mg cholesterol, 11 g fat
1 oz cooked beef liver = 110 mg cholesterol, 1.4 g fat
Supplementation:
Cholesterol supplementation goals:
Infancy: 30-40 mg/kg/day; Older individuals: 50-500 mg/kg/day
Cholesterol supplements available in powder or encapsulated crystalized formulations
Vitamin and mineral supplements: meet DRI, consider ADEK supplementation as needed
Antioxidants: clinical trial suggests evidence for potential benefit[4]

Monitoring

Laboratory[5]
Cholesterol- plasma cholesterol may not reflect tissue levels
Serum concentration of 7-DHC
Serum amino transferases (ALT and AST)
Fat soluble vitamins (A, D, E, K)
Anthropometric
Utilize SLO specific growth charts[3]

Supportive Needs for Feeding

Common symptoms impacting nutrition: Cleft palate, constipation, gastroesophageal reflux, hypotonia, poor feeding/sucking

Address constipation: assess and support fluid needs, encourage intake of insoluble fiber
Support feeding challenges: feeding therapy, g-tube, behavior modification

References

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2. Elias, ER; et al. Cholic acid increases plasma cholesterol in Smith-Lemli-Opitz syndrome: A pilot study. Mol Gen Metab Rep. 2023 Nov 28;38:101031
3. Lee, RWY; et al. Growth charts for individuals with Smith-Lemli-Opitz syndrome. Am J Med Genet A. 2012 Nov;158A(11):2707-13 2012
4. Fliesler, SJ; Antioxidants: The Missing Key to Improved Therapeutic Intervention in Simth-Lemli-Opitz Syndrome? Hereditary Geneti 2013. 2: 119. doi: 10.4172/2161-1041.1000119
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