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# Main clinical characteristics of Smith-Lemli-Opitz syndrome

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## ABSTRACT

Smith-Lemli-Opitz syndrome (SLOS) results from mutations in the DHCR7 gene (11q13), causing cholesterol deficiency and 7-dehydrocholesterol accumulation in the tissues. This causes a variety of symptoms, such as intellectual disability and craniofacial malformations.

**Keywords**: Smith-Lemli-Opitz syndrome, Inborn errors of lipid metabolism, Congenital abnormalities.

## OBJECTIVES

Investigate the main clinical manifestations of Smith-Lemli-Opitz Syndrome (SLOS) found in specialized literature, contributing to an in-depth understanding of this syndrome.

## MATERIALS AND METHODS

This literary review study, conducted in February 2024, analyzed articles from the National Library of Medicine (PUBMED) and SciElo. Using Health Sciences Descriptors (known by the acronym DECs) and Medical Subject Headings (known by the acronym MeSH): “Smith-Lemli-Opitz syndrome” and “clinical manifestations”, theses and reviews were excluded. Reading is based on titles and abstracts, prioritizing research published in the last 5 years in Portuguese, English and Spanish, ensuring relevance and timeliness in the study.

## RESULTS AND DISCUSSION

SLOS presents a wide range of clinical manifestations, highlighted by intellectual disability, craniofacial malformations and developmental challenges. It is interesting to note that, even among individuals sharing the same mutation, the expression of symptoms can vary significantly. In the dento-maxillofacial context, distinct characteristics are observed such as crowded teeth, irregular incisor spacing, oligodontia, polydontia, premature tooth eruption, enamel hypoplasia, bifid uvula, wide alveolar crests and symptoms of Pierre-Robin syndrome, such as glossoptosis, retrognathia and cleft palate.

In addition to facial aspects, SLOS is also associated with neurological manifestations, including microcephaly and holoprosencephaly, in addition to marked intellectual impairment and behavioral challenges. Anthropometric abnormalities, with emphasis on growth restriction, are common physical

manifestations of this syndrome. This complexity and variability in symptom expression reinforce the genetic uniqueness of SLOS.

## CONCLUSION

In summary, SLOS stands out for the diversity of clinical manifestations, demanding a personalized and multidisciplinary clinical approach. The genetic uniqueness of this condition highlights the continued need for research to improve diagnostics and therapeutic strategies.

Therefore, an in-depth understanding of the clinical implications of SLOS is essential to provide comprehensive care and improve the quality of life of affected individuals, reinforcing the importance of collaboration between different medical specialties.

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