

# LEMLI OPITZ SYNDROME: A CASE REPORT

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## ABSTRACT

A case of Smith-Lemli-Opitz syndrome is presented. One-year old child presented with febrile illness and was malnourished with all the three-growth parameters below the 3rd percentile. He was blind since birth and had characteristic facial feature with broad nasal tip anteverted nostrils, micro-opthalmia, rowing eye movements, strabismus, epicanthic folds, long philtrum, low set ears, thin upper lip and oligodontia. Investigations showed cardiac abnormalities and serum cholesterol was 60mg/dl.

**KEY WORDS:** Smith-Lemli-Opitz syndrome, 7-dehydrocholesterol-delta 7-reductase, Congenital Anomalies

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## INTRODUCTION

Smith-Lemli-Opitz syndrome (SLOS) is an autosomal recessive genetic condition caused by deficiency of the enzyme 7-dehydrocholesterol-delta 7-reductase. This enzyme converts 7-dehydrocholesterol (7 DHC) to cholesterol resulting in generalized cholesterol deficiency.<sup>1,2</sup> Cases of SLOS can vary widely in their clinical presentation. The most commonly observed features include characteristic dysmorphic features, syndactyly, growth retardation, microcephaly, intellectual disability and ambiguous genitalia.<sup>3,4</sup>

## CASE HISTORY

A one-year old child was brought to out patient department by his parents with the complaints of fever, cough and loose motion since six days. Examination reveals malnourished child weighing 5.5 kg, his total length was 59 cms and occipital frontal circumference (OFC) was 34cms, all the three-growth parameters were below the 3rd percentile. He had

characteristic facial features (Figure 1) with broad nasal tip anteverted nostrils, micro-opthalmia, rowing eye movements, strabismus, epicanthic folds, long philtrum, low set ears, thin upper lip and oligodontia.

Regarding systemic examination, auscultation of heart revealed ejection systolic murmur best heard at left upper sternal border while auscultation of chest revealed bilateral equal air entry with occasional rhonchi. Tone was decreased in all four limbs but reflexes were normal. Genitalia examination showed underdeveloped scrotum with microphallus and bilateral undescended testis. Regarding the development, all the four area of development were delayed with head holding achieved at the age of 11 months, palmer grasp was still present and showed social smile on tactile stimulation, He could utters vowels but unable to say singleword. He was blind since birth so vision was not assessed. He was born full term at home with no history of cyanosis or delayed cry. He was

second issue of consanguineous marriage with history of four sibling's death (2 still births and 2 intrauterine deaths).

His routine laboratory investigations like complete blood picture, renal function tests, urine R/E were normal. His serum cholesterol was 60mg/dl, ultrasound abdomen was normal but ultrasound scrotum showed hypo plasticsac with no evidence of testes in scrotum. Left small ectopic testicle was in left inguinal region but the right testicle was not identified in inguinal region or pelvis. Echocardiography showed atrial septal defect with left to right shunt and moderate pulmonary stenosis while CT brain showed mild brain atrophy (Figure 2).

## DISCUSSION

Smith-Lemli-Opitz syndrome (SLOS) is a rare hereditary disease characterized by prenatal and postnatal growth retardation, microcephaly, and variable degree of intellectual disability with multiple organ involvement. All these features lead to significant morbidity and poor quality of life not only for the child but also for the entire family.

As SLOS is very rare in Asia and relatively more frequently reported from west,<sup>5</sup> so we are probably the first to report this case in national literature.

The weakness of this case report is that we have diagnosed this child on the basis of clinical examination and supported biochemically by low cholesterol level however the definite diagnosis is based on high level of 7-dehydrocholesterol (7 DHC) in serum or tissues but this diagnostic marker is unfortunately not available in Pakistan.



Figure 1: Characteristic facial and limb features of Smith-Lemli-Opitz syndrome

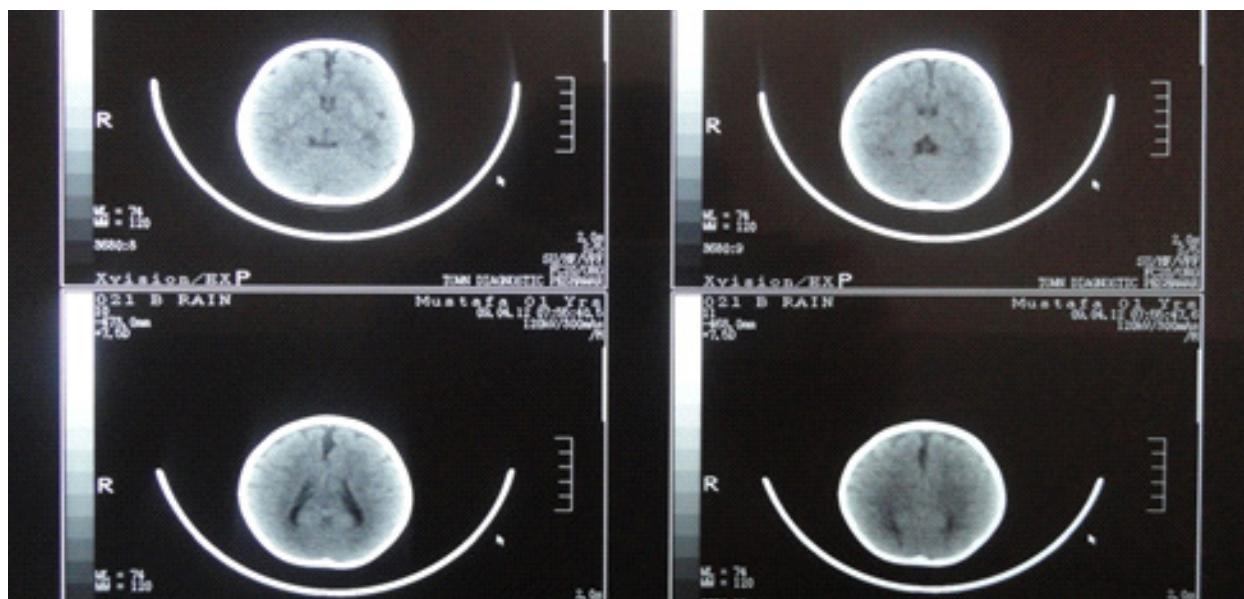


Figure 2: CT Brain showing mild cerebral atrophy

It was first described in 1964 by the late David Smith, the Belgian pediatrician Luc Lemli, and John Opitz.<sup>6</sup> In the majority of case report published, smith Lemli Opitz was diagnosed within first year of life and this was also observed in our report. SLOS is mostly diagnosed clinically with typical dysmorphic features, microcephaly, mental retardation, congenital heart defect and ambiguous genitalia and the same features were also present in our case.<sup>7</sup>

Infants with SLOS are almost always small for gestational age and most con-

tinue to grow below the 3rd centile. This finding is also observed in our case too.<sup>8</sup>

Although the diagnostic test is the elevated level of 7-dehydrocholesterol (7DHC) in serum or tissues but most of authors have stressed that it can be easily diagnosed clinically and biochemically by low cholesterol level.<sup>9</sup> The case report from India did not mention the diagnostic marker in their study probably due to non-availability of this test.<sup>10</sup> The same was observed in our case report, as this is not available in the best laboratory of Pakistan.

Our case report benefits the neonatologist, pediatrician, pediatric plastic surgeon, endocrinologist and urologist.

## CONCLUSION

SLOS is a rare disease presenting as multiple congenital anomalies. The typical features are syndactyly, congenital heart disease, ambiguous genitalia and mental retardation. so any child having these findings should be investigated for Smith Lemli Opitz syndrome.

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### CONFLICT OF INTEREST

Authors declare no conflict of interest

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