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 antverted nostrils, micro-ophtalmia, rowing eye movements, strabismus, epicanthic folds, long philtrum, low set ears, thin upper lip and oligodentia. Investigations showed cardiac abnormalities and serum cholesterol was 60mg/dl.

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

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.1,2 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.3,4

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 antverted nostrils, micro-ophtalmia, rowing eye movements, strabismus, epicanthic folds, long philtrum, low set ears, thin upper lip and oligodentia.

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 descended testes. 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 unable to say single word at home. 

His routine laboratory investigations 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,5 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.

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

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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.
REFERENCES


CONFLICT OF INTEREST
Authors declare no conflict of interest

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