Smith-Lemli-Opitz Syndrome

A 15-month-old child was brought for evaluation of ambiguous genitalia and failure to thrive. He was irritable, underweight and short for age. He had hypopigmented hair, hypertonia, scissoring of lower limbs, asymmetrically short flexed fingers, microcephaly, narrow frontal area, slanting auricles, broad nasal tip, anteverted nostrils, micrognathia, cleft palate, thickened dental ridge, simian crease, polydactyly, metatarsus abductus, syndactyly of second and third toes, and ambiguous genitalia (micropenis, hypospadias, cryptorchidism and a rudimentary vagina with a karyotype of 46XY) (*Fig. 1*). The child had pyloroplasty for congenital hypertrophic pyloric stenosis at 28 days of age. An ultrasound examination of abdomen revealed left sided hydronephrosis, duplex collecting system, ureterocele and cystic dysplasia of upper calyceal system. His lipid profile revealed low serum cholesterol levels and absent LDL cholesterol. A diagnosis of Smith-Lemli-Opitz syndrome was made based on the clinical and biochemical profile.

Smith-Lemli-Opitz syndrome, first described in 1964, is a rare autosomal recessive disorder of cholesterol synthesis wherein the conversion of 7-dehydro-cholesterol (7-DHC) into cholesterol is disrupted leading to excessive accumulation of 7-dehydro-cholesterol, 8-dehydro-cholesterol (8-DHC, an isomeric form of 7-DHC), and deficiency of cholesterol. The accumulated 7-DHC and 8-DHC also cause decreased HMG-CoA reductase activity. Hence cholesterol replacement forms the cornerstone in medical management of this condition. The coexisting surgical conditions may need appropriate evaluation and intervention.

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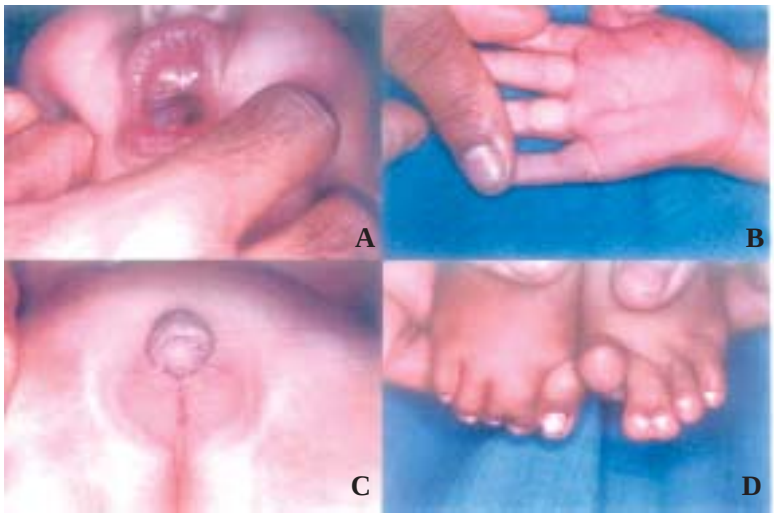

Fig. 1. A. Showing cleft palate and thickened alveolar ridges, B. showing polydactyly and simian crease. C. showing ambiguous genitalia and D. showing syndactyly of second and third toes.