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Smith Lemli Opitz Syndrome: A Disease Process To Look Out For?

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Smith Lemli Opitz syndrome [aka SLOS] is a genetic disorder, which can be manifested by a number of physical characteristics, primarily this is a disorder of cholesterol metabolism1. This disease process has a broad spectrum of phenotype from a mild manifestation to life-threatening syndrome[2].

In this syndrome, cholesterol level is decreased and dehydrocholesterol level is increased due to mutations of 7-dehydrocholesterol reductase [7DHCR][2]. Although cholesterol level doesn't act as an indicator of this disease, diagnosis can be confirmed by finding elevated 7DHCR, in plasma or tissues[2]. Combination of low cholesterol and high 7DHC suggests a block in cholesterol biosynthesis route[3].

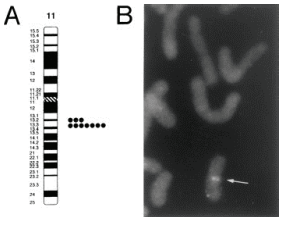

Fig: localization of human 7-DHCR gene4.

Physical and clinical manifestation of SLOS can range from growth retardation, developmental delay, microcephaly, cleft palate, syndactyly of the 2nd and 3rd toes, dry skin to systemic defects such as atrial/ ventricular septal defects, patent ductus arteriosus, hypertension, constipation, pyloric stenosis, malrotation, hypospadias, cryptorchidism, hypotonia, autism etc.2

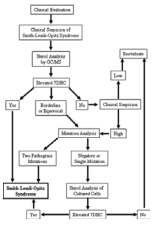

Figure: Flowchart to diagnose SLOS2

In a surgical context, patients presenting with ambiguous genitalia, Hirshprung's disease and pyloric stenosis, may trigger screening of SLOS, leading to diagnosis[5].

Cholesterol supplements are the mainstay of treatment. However, they can treat brain defects as cholesterol cant cross blood-brain barrier [BBB]2. Simvastatin therapy shows more promises[2].

Prenatal diagnosis is possible by either amniotic fluid or chorionic villus sampling

Biography

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