7 - Dehydrocholesterol

Description	7-dehydrocholesterol is a cholesterol precursor found in the skin. It is converted to cholecalciferol (vitamin D3) in the presence of ultraviolet light. 7-dehydrocholesterol is used as a biochemical marker for the diagnosis of Smith-Lemli-Opitz Syndrome.
Indication	Diagnosis of Smith-Lemli-Opitz Syndrome.
Additional Info	Smith-Lemli-Opitz Syndrome is a condition with multiple congenital malformations. It is caused by mutations in the DHCR7 gene leading to deficient activity of 7-dehydrocholesterol reductase, the final enzyme of the cholesterol biosynthetic pathway. Since 7-dehydrocholesterol reductase drives the conversion of 7-dehydrocholesterol to cholesterol, deficiency of this enzyme results in low cholesterol levels (an essential component of cell membranes) and high concentrations of its direct precursor 7-dehydrocholesterol in body fluids and tissues. The enzyme defect also causes reduced myelination in the cerebral hemispheres, cranial nerves and peripheral nerves. The characteristic features observed in Smith-Lemli-Opitz Syndrome include microcephaly, anteverted nares, micrognathia, syndactyly of the 2nd and 3rd toes, genital anomalies, severe learning difficulties, behavioural problems and tendency to self-injury. Treatment options include a combination of cholesterol supplementation and statin therapy (to reduce the build-up of 7-dehydrocholesterol).
Concurrent Tests	Prenatal testing: Amniocentesis at ~15 weeks gestation.
Dietary Requirements	N/A
Interpretation	For reference interval see report or contact laboratory Prenatal testing: Amniotic fluid analysis by gas chromatography-mass spectrometry demonstrates a low cholesterol concentration and markedly elevated 7-dehydrocholesterol concentration.
Collection Conditions	N/A
Frequency of testing	As required.