Memo No.   2021-019

Date: 24-Jun-2021

Memo To: Clients

Re: *Test Revision - 7-Dehydrocholesterol, Serum/Plasma*

7-dehydrocholesterol (7DHC) is the biomarker for the diagnosis of the cholesterol biosynthesis defect Smith-Lemli-Opitz syndrome (SLOS). Effective June 15, the test methodology for 7-dehydrocholesterol measurement was revised to a new, more sensitive liquid chromatography-mass spectrometry (LCMS) method. Previously, the method was not sensitive enough to quantify 7DHC in the normal range, but the new method has reference intervals for the normal population (see table). Additionally, **Amniotic Fluid samples will no longer be accepted for analysis**.

As part of this change, additional cholesterol biosynthesis intermediates and sitosterol are also going to be measured (see table for full list, including reference intervals). Therefore, the test name is being revised to Sterol Profile, Serum/Plasma. 7-Dehydrocholesterol, Amniotic Fluid has been deleted from the online catalogue.

STEROL PROFILE	REFERENCE INTERVAL	CLINICAL INDICATION
Analyte	**umol/L**	**(elevation)**
7-dehydrocholesterol	(0.3 - 2.2)	SLOS
8-dehydrocholesterol	(0.2 - 3.7)	SLOS
cholestanol	(0.0 - 8.6)	CTX
desmosterol	(0.5 - 6.7)	desmosterolosis
lanosterol	(0.0 - 0.7)	Antley-Bixler syndrome-1
lathosterol	(0.0 - 0.0)	lathosterolosis
sitosterol	(1.7 - 34.7)	sitosterolemia

Notes: Cholestanol is 5α-cholestan-3β-ol. CTX is cerebrotendenous xanthomatosis. Lanosterol *may* be elevated in Antley-Bixler syndrome-1. Cholesterol biosynthesis disorders present with multiple congenital anomalies (most) or neurological symptoms with xanthomas (CTX). Sitosterol is elevated in sitosterolemia, which presents with xanthomas and accelerated atherosclerosis.

If you have further questions, please contact Client Care at (416) 422-3000 Ext. 300 or info@iclabs.ca

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