

## CHOLESTANOL

### Relevant disorders

Cerebrotendinous Xanthomatosis

### Related Metabolic Tests

Bile acids (urine)

### Indication for Test

Cholestanol. (5 $\alpha$ -cholestan-3 $\beta$ -ol or Dihydrocholesterol) is formed from cholesterol after four enzymatic transformations. This compound is significantly increased in the plasma of patients with the disorder Cerebrotendinous xanthomatosis (CTX) (mitochondrial sterol C27 hydroxylase deficiency). Normally metabolised to bile acids, the build up of cholestanol in tendon and neural tissues causes death from the fourth decade and can be associated with high plasma cholesterol. Clinical observations include psychomotor retardation, xanthomas, dementia, cataract and atherosclerosis.

### Methodology

Any esterified plasma sterols are hydrolysed by alkali in ethanol at 55°C. The free sterols are then extracted into heptane and after drying are converted into trimethylsilyl (TMS) ethers for analysis by GCMS. An analogue internal standard (coprostanol or 5 $\beta$ -cholestan-3 $\beta$ -ol) is used for quantitation. Calibration curves are constructed for cholesterol, cholestanol and 7-dehydrocholesterol – standards are run with each assay. A separate qualitative analysis is used to detect any other abnormal plasma sterols such as desmosterol, lathosterol, and lanosterol.

### Sample requirements

1 ml Lithium heparin blood, separate and send plasma (Serum acceptable).

## Transport information/Contact details

Send all samples by first class post to:

Department of Clinical Chemistry  
Sheffield Children's NHS Foundation Trust  
Western Bank, Sheffield  
S10 2TH, UK

Joanne Croft (Clinical Scientist)  
0114 2717307

## Turn Around Time

4 weeks

## Reference Ranges

Normal range = 3 – 16  $\mu\text{mol/L}$

N.B: these reference ranges are only valid with data produced by the Department of Clinical Chemistry, Sheffield Children's NHS Foundation Trust.

## References

- The Metabolic and Molecular Bases of Inherited Disease Eighth edition 2001 pages 2970 – 2977. Scriver et al Eds.McGraw-Hill