8 DEHYDROCHOLESTEROL

Relevant disorders

Conradi – Happle - Hunermann syndrome (aka X-linked dominant chodrodysplasia punctata or Eopamil binding protein deficiency)

Indication for Test

Females: rhizomelic short stature with aysymmetric shortening of the proximal limbs, stippled epiphyses, sectorial cataracts, intellectual disability

Males: lethal

Methodology

Any esterified plasma sterols are hydrolysed by alkali in ethanol at 55^{o}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 5B-cholestan-3B-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.

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 (but if requested urgently can be performed more quickly)

Reference Ranges

Interpretation will be provided with the report.

References

- Kelley and Kratz Paed. Res. 37.No5.671-675.1995
- Smith DW et al A newly recognised syndrome of multiple congenital anomalies (1964) J.Pediatrics Vol 64 no.2 210-217
- Clayton PT Disorders of cholesterol biosynthesis (1998) Arch Dis Child 78: 185-189
- Jira PE et al Pitfalls in measuring plasma cholesterol in the Smith-Lemli-Opitz syndrome. (1997) Clinical Chemistry 43:1 129-133