

## PHYTOSTEROLS

### Relevant disorders

Phytosterolaemia

### Indication for Test

Phytosterolaemia is an inherited metabolic disorder affecting the transport of plant derived sterols from the gut by the ATP-binding cassette transporter Sterolin. Loss of selectivity of the transporter results in increased circulating plasma phytosterols (mainly sitosterol). These sterols cannot be metabolised and thus accumulate in tissues causing xanthomata and atherogenicity, as well as haematological symptoms associated with thrombocytopenia and stomatocytosis. In fact, there is probably a discrete group of patients who have no xanthomata but growth retardation and haematological symptoms. Treatment has been limited to dietary restriction of phytosterols (e.g. low soya or olive oil), although some new sterol absorption blocker compounds have been proposed for this condition.

### 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 5B-cholestan-3B-ol) is used for quantitation.

### 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

Interpretation will be provided with the report.

## References

- The Metabolic and Molecular Bases of Inherited Disease Eighth edition 2001 pages 2970 – 2977. Scriver et al Eds.McGraw-Hill
- Stomatocytic haemolysis and macrothrombocytopenia (Mediterranean stomatocytosis/macrothrombocytopenia) is the haematological presentation of phytosterolaemia.  
Br J Haematol. 2005 Jul;130(2):297-309. Rees DC, Iolascon A, Carella M, O'marcaigh AS, Kendra JR, Jowitt SN, Wales JK, Vora A, Makris M, Manning N, Nicolaou A, Fisher J, Mann A, Machin SJ, Clayton PT, Gasparini P, Stewart GW.