Phytanic Acid

Description	3-methyl fatty acid (Included in very long chain fatty acid screen)
Indication	Investigation of suspected disorders of peroxisomal metabolism.
Additional Info	Phytanic acid undergoes alpha-oxidation in peroxisomes. Clinical features of inborn errors of peroxisome metabolism include neurological, skeletal, ocular, craniofacial and hepatointestinal abnormalities. Plasma very long chain fatty acid analysis is the most useful screening test for these conditions.
Concurrent Tests	Very long chain fatty acids (VLCFA) Pristanic acid Plasmalogens Dihydroxyacetonephosphate acyltransferase
Dietary Requirements	N/A
Interpretation	Phytanic acid is typically increased in disorders of peroxisome biogenesis, Refsum disease and rhizomelic chondrodysplasia punctata, but is normal in X-linked adrenoleukodystrophy. Phytanic acid levels will only be abnormally raised after sufficient dietary intake, i.e. older patients. Fibroblast assays may be required to confirm the diagnosis.
Collection Conditions	Avoid sending to laboratory on Friday. All relevant clinical information should be provided with the sample. 0.5 mL lithium heparin plasma, 1st class post, Sheffield Children's hospital or to send sample to Willink (Manchester): can do all tests on same 5 mL sample.
Frequency of testing	Repeated measurement inappropriate