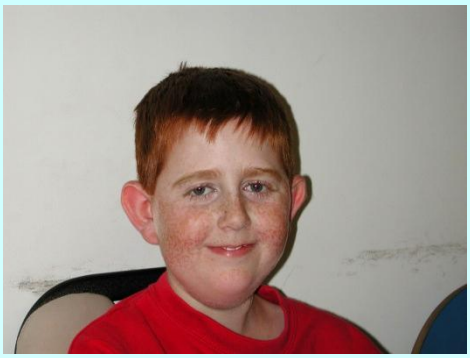
# National Barth Syndrome Service

**An NHS Specialised Service for Barth Syndrome commenced April 2010 at Bristol Royal Hospital for Children.** Biochemical and genetic testing performed in Bristol at no charge to users. Retrospective diagnosis should be considered in all cases of male DCM, even if this has completely resolved.

**Diagnosis**: elevated ratio of MLCL/CL (monolysocardiolipin / cardiolipin) on a 3ml EDTA blood sample, or blood filter paper spots, stored Guthrie spots, fibroblasts or tissue.

## Consider Barth Syndrome in patients with any of the following features:

**FAMILY HISTORY: unexplained fetal cardiomyopathy, third trimester loss, stillbirth, family history of child male death (sudden / infective / cardiac)**

- **CARDIAC dilated cardiomyopathy +/- endocardial fibroelastosis, left ventricular compaction**
- **MOTOR PROBLEMS: delayed motor milestones, proximal myopathy, fatigue**
- **DYSMORPHIC FEATURES : deep set eyes, prominent ears, tall forehead**
- **HYPOGLYCAEMIA / LACTIC ACIDOSIS especially in the neonatal period or infancy**
- **GROWTH RETARDATION during childhood; rapid growth in late adolescence**
- **NEUTROPAENIA: mild to severe; intermittent, cyclical or persistent; may be absent**
- **GUT PROBLEMS: Feeding problems, vomiting, recurrent diarrhoea, savoury food fads**
- **ABNORMAL MITOCHONDRIA or respiratory chain tests**
- **UNEXPLAINED VENTRICULAR ARRHYTHMIA or SUDDEN DEATH**

Further advice about free testing and the service are available at:
***www.barthsyndromeservice.nhs.uk*** ***or*** ***www.barthsyndrome.org.uk***

for further information please contact:
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