

# Could he have Barth Syndrome?



**BARTH SYNDROME** is an under-recognised cause of cardiac problems in boys, causing 3-5% of dilated cardiomyopathy (DCM) in a large American study.

**An NHS Specialised Service at Bristol Royal Hospital for Children commenced in April 2010.**

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.

**Barth Syndrome should be excluded in patients with any of the following cardiac features:**

- **DILATED CARDIOMYOPATHY +/- ENDOCARDIAL FIBROELASTOSIS**
- **LEFT VENTRICULAR NON-COMPACTION**

In all **NEONATAL** or **INFANT** cases plus older boys with one or more of the following:

- **SUSPICIOUS FAMILY HISTORY:** Fetal cardiomyopathy, third trimester loss, stillbirth, family history of child male death (sudden / infective / cardiac)
- **MOTOR PROBLEMS:** delayed motor milestones, proximal myopathy, fatigue
- **CHARACTERISTIC FACIAL APPEARANCE:** deep set eyes, prominent ears
- **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
- **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](http://www.barthsyndromeservice.nhs.uk) or [www.barthsyndrome.org.uk](http://www.barthsyndrome.org.uk)

or Debbie Riddiford, Clinical Nurse Specialist on 07795 507294  
or for enquiries about testing Ms Ann Bowron on 0117 342 2590