

Could he have Barth syndrome?



BARTH SYNDROME is an under-recognised cause of cardiac problems in boys, causing 7% of dilated cardiomyopathy (DCM) in a large American study.

NCG Specialised Service 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 DCM, even if completely resolved.

Diagnosis: elevated ratio of MLCL/CL (monolysocardiolipin / cardiolipin) in tissue, white blood cells, blood filter paper spots or stored Guthrie spots.

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

- **DILATED CARDIOMYOPATHY +/- ENDOCARDIAL FIBROELASTOSIS**
- **LEFT VENTRICULAR NON-COMPACTION or**
- **UNEXPLAINED VENTRICULAR ARRHYTHMIA or SUDDEN DEATH**

together with any 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
- **GI PROBLEMS:** Feeding problems, vomiting, recurrent diarrhoea, savoury food fads
- **ABNORMAL MITOCHONDRIA or respiratory chain tests**

Further advice about free testing and the service are available at:

barthsyndromeservice@UHBristol.nhs.uk or www.barthsyndrome.org.uk

Or by telephone from Debbie Riddiford, Barth Service CNS, on 07795 507294 or Susan Western, Service Administrator, on 07767 310452