National Barth Syndrome Service

Barth Syndrome is X-linked and under-diagnosed.
Please consider in males with any of the following features:

CARDIAC: dilated cardiomyopathy +/- endocardial fibroelastosis (EFE), left ventricular compaction (LVNC), fetal cardiomyopathy, second & third trimester loss, stillbirth, ventricular arrhythmia, family history of fetal & child male death

MYOPATHY: Proximal myopathy, delayed milestones, feeding problems, diarrhoea

UNEXPLAINED "mitochondrial" disease; neonatal/infantile hypoglycaemia and lactic acidosis, abnormal respiratory chain function tests or mitochondrial appearance, 3-methylglutaconic aciduria

DYSMORPHIC FEATURES : deep set eyes, prominent ears, growth retardation during childhood, delayed bone age, rapid growth in late adolescence

NEUTROPAENIA: highly variable within & between patients; mild to severe; intermittent, cyclical or persistent but may be completely absent. MANY SERIOUS OR FATAL BACTERIAL INFECTIONS REPORTED.

An NHS Specialised Service for Barth Syndrome commenced April 2010 at Bristol Royal Hospital for Children. Biochemical (cardiolipin analysis) and genetic (TAZ gene) testing is performed in Bristol free of charge to users. Retrospective diagnosis is recommended in all cases of idiopathic male DCM.

Diagnosis: Cardiolipin profiling is best performed on a 3ml EDTA blood sample. However, blood filter paper spots, such as stored Guthrie spots OR stored fibroblasts and many other tissues can be tested.

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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