Mitochondrial testing m.1555A>G

Introduction

Some mitochondrial point mutations have been associated with deafness, the most commonly reported being m.1555A>G. The homoplasmic mutation m.1555A>G in the mitochondrial MT-RNR1 (12S rRNA) gene has been associated with aminoglycoside-induced and nonsyndromic sensorineural deafness (Estivill X et al, Am J Hum Genet 62(1): 27-35, 1998; Prezant TR et al., Nat Genet 4 (3): 289-294, 1993). The mutation has been detected in families with maternally transmitted deafness and seems to have an age dependent penetrance for deafness, which is enhanced by treatment with aminoglycosides.

Referrals

- Patients with hearing loss for m.1555A>G mutation analysis.
- Patients who may require aminoglycosides.
- Maternal relatives of patients with the m.1555A>G mutation.

Service offered

Mutation analysis for the m.1555A>G mutation.

Technical

Restriction enzyme assay is performed to detect the m.1555A>G mutation. All mutation positive results are confirmed by sequence analysis.

Target reporting time

Two weeks for routine testing of m.1555A>G mutation in index case.
Two weeks for maternal relatives of patients with the m.1555A>G mutation.
Please contact the laboratory for urgent cases.

Contact details

Molecular Genetics Service
Level 6, York House
37 Queen Square
London, WC1N 3BH
T +44 (0) 20 7762 8888
F +44 (0) 20 7813 8578

Samples required

- 5ml venous blood in plastic EDTA bottles (>1ml from neonates)
- Prenatal testing must be arranged in advance, through a Clinical Genetics department if possible.
- Amniotic fluid or CV samples should be sent to Cytogenetics for dissecting and culturing, with instructions to forward the sample to the Regional Molecular Genetics laboratory for analysis
- A completed DNA request card should accompany all samples

Patient details

To facilitate accurate testing and reporting please provide patient demographic details (full name, date of birth, address and ethnic origin), details of any relevant family history and full contact details for the referring clinician