Early-Onset Primary Dystonia (DYT1)/ TOR1A Gene

Clinical Background and Genetics
- DYT1 (Early-onset Primary Dystonia; Torsion Dystonia 1; OMIM: 605204) is a movement disorder characterised by involuntary, repetitive, sustained muscle contractions or postures involving one or more sites of the body.
- The average age of onset is 12 years and in the vast majority of patients onset is before 26 years.
- Posturing or twisting of a foot, leg or arm are the most common presenting findings and may be apparent with specific actions e.g. writing or walking.
- Over time the contractions may progress from a limb to other body regions (generalised dystonia) and become less action-specific.
- The distribution and severity of symptoms can vary widely between affected individuals and within families ranging from mild focal dystonia e.g. writer’s cramp to severe generalised dystonia.
- >99% of individuals with DYT1 have a three base-pair deletion in exon 5 of the TOR1A gene: c.907_909delGAG (p.Glu303del).
- Inheritance is autosomal dominant with reduced penetrance (approx. 30%).

Service offered
- Fluorescent PCR and fragment analysis to detect the common c.907_909delGAG deletion.
- If the deletion is indicated by PCR, confirmatory sequence analysis of exon 5 of the TOR1A gene is performed to confirm the presence of the deletion.

Referrals
- DYT1 genetic testing is recommended for patients with a limb-onset, primary dystonia with onset before age 30. Patients with onset after age 30 may be tested if they have an affected relative with early-onset dystonia.
- DYT1 testing is not recommended in asymptomatic family members due to reduced penetrance of the disease.
- Referrals accepted from Clinical Genetics, Paediatrics, Neurology

Target reporting Time
Diagnostic testing: 20 day turn-around time

For up-to-date prices please contact the laboratory

Clinical Advice: If clinical discussion is required we would recommend discussion with Dr Peter Lunt, Consultant Clinical Geneticist, St Michael’s Hospital, Bristol (Tel: 0117 342 5652).