INTRODUCTION
X-linked Charcot-Marie-Tooth is a hereditary or genetic neuropathy that is inherited in an X-linked dominant manner. It represents at least 10-20% of hereditary neuropathies and is characterised by moderate to severe peripheral motor and sensory neuropathy in affected males. The phenotype in carrier females is variable, ranging from having no or mild symptoms to having pronounced symptoms due to skewed X-inactivation. Sensorineural deafness and central nervous system symptoms also occur in some families.

CMTX is associated with mutations in the \textit{GJB1} (\textit{Cx32}) gene located at Xq13.1.

TESTING AND REFERRALS
Diagnostic:
- Clinically affected patients
- From Clinical Genetics, Clinical Neurology, Paediatrics and Paediatric Neurology

Presymptomatic:
- Referrals for asymptomatic individuals with clinically affected relatives
- From Clinical Genetics only

Prenatal:
- Available to couples at risk of having a child affected with CMTX
- Referrals are only accepted from Clinical Genetics and where possible, arranged in advance

STRATEGY AND TECHNICAL INFORMATION
- Mutation screening in diagnostic referrals is undertaken by bidirectional fluorescent sequencing of the coding region (exon 2) of the \textit{GJB1} (previously \textit{Cx32}) gene.
- When a pathogenic mutation has been identified in an individual, subsequent testing of family members (presymptomatic) involves testing for the familial mutation only.

TARGET REPORTING TIMES
Diagnostic (\textit{GJB1} sequencing): 40 days
Presymptomatic: 10 days
Prenatal testing (includes maternal contamination check): 3 days

N.B Details are correct for the date of printing only – last updated 08/05/2015