CONGENITAL MYASTHENIC SYNDROME (CMS)

OMIM 100690 / 100710 / 100720 / 100725 / 100730 / 603033 / 118490 / 601592 / 610285 / 191350 / 138292

INTRODUCTION
Congenital Myasthenic Syndrome (CMS) is caused by defective transmission at the neuromuscular junction and results in fatigable muscle weakness. CMS is a clinically and genetically heterogeneous condition. Inheritance is usually autosomal recessive but can be autosomal dominant. Mutation screening is available for eleven genes.

TESTING - All samples MUST be accompanied by a completed Pre-referral form (click here)
- Diagnostic: Clinically affected patients
- Carrier/Presymptomatic/Familial mutation test: Relatives of clinically affected patients (known mutation)
- Prenatal: At risk of having an affected child (known mutation)

REFERRALS
- From Clinical Genetics, Clinical Neurology and Paediatric Neurology
- Prenatal referrals are only accepted from Clinical Genetics and/or Prenatal Diagnosis. They must be discussed with the laboratory and arranged in advance.

STRATEGY
- Clinical review with targeted gene screening determined by the patient’s clinical phenotype, family history and ethnic background

TECHNICAL INFORMATION
- Sequencing analysis of exons 1-9 and splice site boundaries of the CHRNA1 (αAChR-subunit) gene
- Sequencing analysis of exons 1-11 and splice site boundaries of the CHRNB1 (βAChR-subunit) gene
- Sequencing analysis of exons 1-12 and splice site boundaries of the CHRND (δAChR-subunit) gene
- Sequencing analysis of the N-box, exons 1-12 and splice site boundaries of the CHRNE (εAChR-subunit) gene
- Sequencing analysis of exons 1-17 and splice site boundaries of the COLQ gene
- Sequencing analysis of exons 5-18 and splice site boundaries of the CHAT gene
- Sequencing analysis of the E-box, exons 1-8 and splice site boundaries of the RAPSN gene
- Sequencing analysis of exons 1-7 and splice site boundaries of the DOK7 gene
- Sequencing analysis of exons 1-9 and splice site boundaries of the DPAGT1 gene
- Sequencing analysis of exons 1-20 and splice site boundaries of the GFPT1 gene

TARGET REPORTING TIMES
- Diagnostic test: 40 days
- Carrier/Presymptomatic/Familial mutation test: 10 days
- Prenatal test (includes maternal contamination check): 3 days

This service is funded through the Highly Specialised Services (HSS) for patients living in England and Scotland.

N.B. Details are correct for the date of printing only – last updated 18/08/2015