GLUT1 DEFICIENCY SYNDROME
- OMIM 138140 (SLC2A1)

INTRODUCTION
SLC2A1 (also known as GLUT1) encodes the glucose transporter 1 protein (GLUT1), and is the only gene known to be associated with GLUT1 deficiency syndromes. GLUT1 deficiency syndromes are a group of autosomal dominant neurological disorders with hypoglycorrhachia (low CSF glucose) and wide phenotypic variability (OMIM #606777, #601042, #612126, #614847), most commonly including epilepsy and movement disorder. The majority of cases are sporadic due to de novo mutations. A small number of rare cases with autosomal recessive inheritance have been reported.

SLC2A1 analysis is available as a specific single gene service as summarised below, or alternatively is included as part of a panel of 98 genes associated with ataxia (refer to separate ataxia service page for details).

TESTING
- Diagnostic: Clinically affected patients
- Carrier or Presymptomatic: Relatives of clinically affected patients
- Prenatal: At risk of having an affected child

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

STRATEGY
- Sequencing of the coding region of SLC2A1 and dosage analysis to test for exonic deletions/duplications.

TECHNICAL INFORMATION
- Sanger sequencing of the exons 1-10 of SLC2A1.
- Dosage analysis of all exons of SLC2A1 by MLPA using kit P138 from MRC-Holland.

TARGET REPORTING TIMES
- High priority diagnostic tests: 10-20 working days
- Routine diagnostic tests: 40 working days
- Carrier/Presymptomatic tests: 10 working days
- Prenatal testing (includes maternal contamination check): 3 working days

N.B. Details are correct for the date of printing only – last updated 04/12/2015