Intact glycoprotein (transferrin) analysis to screen for congenital disorders of glycosylation (CDG) SERUM

NHRPL Tariff Code:
4268
Tariff (Including VAT):
R 3,248.84*
*The listed tariff does not include logistical, collection and other services.
Mnemonic:
PCDGQUANT
Turnaround time:
30 work days from receipt of sample at international laboratory (sendaway test)
Method:
LC-MS/MS glycoprotein intact (transferrin) analysis - Send-away test

Description

Above price includes the assay, quantification and interpretation

Comments

1. This test is utilised in the diagnosis of congential disorders of glycosylation. This include type 1 and to some extent type 2 subtypes. Some CDGs (such as ALG13-CDG) do not present with an abnormal glycoprotein profile.
2. Blood transfusion may influence the analysis. We recommend this test 2 weeks after transfusion to prevent false negative results.

Information Required with sample(s):

Absent clinical details may affect the interpretation of results and recommendations for further/additional testing and subsequent diagnosis of a metabolic disorder. Consent to use below information (point 4) is required according to POPIA regulation:

1. Clinical history of the patient. The referring clinician can complete the clinical history form by downloading it from the website and sending it with the sample, or emailing it to pliem@nwu.ac.za.
2. Other relevant medical reports (e.g. MRI brain, EEG, X-Ray reports, sonar reports, biopsy reports, genetic testing reports, etc) that may assist in the diagnosis of a metabolic disorder can be emailed to pliem@nwu.ac.za.
3. Cumulative, routine pathology results of the patient (including archive results available) – this must be provided and emailed to pliem@nwu.ac.za by the referring pathology laboratory.
4. Please download and complete the short consent form from the website and also indicate if the patient/family may be contacted by our rare disease biobank.

Sample requirements, viability, stability:

1. A minimum of 2 ml SST (yellow top tube) serum is required.
2. Sample should be frozen, and kept frozen during transport.
3. A haemolysed sample is NOT viable for testing as this may lead to the reporting of false positive / negative findings.
4. Viability: 6 Months kept frozen.