NMP NATIONAL METABOLOMICS PLATFORM

Newborn Screening (South Africa)
DRIED BLOOD SPOT

NHRPL Tariff Code:
4083 or (4238 x 3 + 4268 + 4592)
Tariff (Including VAT):
R 1,761.39
Mnemonic:
NNBS
Turnaround time:
10 work days from receipt of sample at CHM.
Method:
Tandem-Mass spectrometry, Fluoro-immunoassay, Enzymatic analysis

Description

Carnitines: Free carnitine (C0), Acetyl (C2)-, Propionyl (C3)-, Butyryl (C4)-, Isovaleryl (C5)-, 3-Hydroxyisovaleryl (C5OH)-, Glutaryl (C5DC)-, Hexanoyl (C6)-, Octanoyl (C8)-, Decanoyl (C10)-, Decenoyl (C10:1)-, Myristoyl (C14)-, Tetradecenoyl (C14:1)-, Palmitoyl (C16)- and 3-Hydroxyhexadecanoyl (C16OH)-carnitine.
Amino acids: Phenylalanine, Leucine/Isoleucine, Valine, Methionine, Citrulline and Tyrosine.
Other: GALT enzyme activity, Biotinidase enzyme activity, Immunoreactive trypsinogen, 17-Hydroxyprogesterone, Thyroid stimulating hormone.

Comments

Primary disorders targeted in the newborn screening program:
Amino Acid Disorders: Classic phenylketonuria, Homocystinuria, Maple syrup urine disease, citrullinemia Type I.
Organic acid disorders: Isovaleric acidemia, Propionic acidemia, Methylmalonic acidemia, Glutaric acidemia type I, Holocarboxylase synthase deficiency, 3-Methylcrotonyl-CoA carboxylase deficiency, 3-Hydroxy-3-methyglutaric aciduria, ß-Ketothiolase deficiency.
Fatty acid oxidation disorders: Medium-chain acyl-CoA dehydrogenase deficiency, Very-long-chain Acyl-CoA dehydrogenase deficiency, Long-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency/Trifunctional protein deficiency, Carnitine uptake defect/Carnitine transport defect.
Other: Classic galactosemia, Congenital adrenal hyperplasia, Primary congenital hypothyroidism, Biotinidase deficiency, Cystic fibrosis.
The primary disorders are diseases for which testing has a high diagnostic accuracy and where early intervention improves outcome. Note however that the test is a screening test. A negative result does not completely exclude the primary disorders. Due to the non-specific nature of metabolic markers, diseases other than the primary disorders, may be identified. Tyrosinemia type I has been removed from the list of primary screening disorders because the diagnostic accuracy of tyrosine was shown to be insufficient. Immunoreactive trypsinogen is an unreliable screening test for cystic fibrosis when meconium ileus is present.

Information Required with sample(s):

Information that is required for the interpretation of the results:

1. Name and surname on both the dried blood card and the requisition form.
2. Date and time of birth.
3. Date and time of sample collection. Note that interpretation of the results requires the age of the patient to the nearest hour at the time of sample collection
4. Weight and gestational age at birth.
5. Gender, Feeding (please indicate the use of lactose free formulations) & Ethnicity
6. Specify the use of maternal steroids, antibiotics, Anticonvulsants, L-Carnitine, TPN and blood transfusions.
7. Specify if the collection is a first or repeat collection.

Absent clinical details may affect the interpretation of results and recommendations.

Sample requirements, viability, stability:

Whatman 903 dried blood spot card with 4 blood spots. Cards must be sent on a separate requisition number and should not include any other requests/samples.
Samples must be collected according to the NBS sample collection standard operating procedure. EDTA samples are not viable for NBS testing. Air dry collected samples for at least two hours before placing in a sealed paper envelope. Collected samples should be kept at room temperature. Avoid high humidity and temperatures. Samples must reach the CHM within two days from collection.