Protocol for newborn screening

General

A newborn screening for inborn metabolic diseases is widely done by tandem-MS or GC-MS measurements. Both techniques can only determine 40 metabolites, while $^1$H-NMR spectroscopic investigations of urine samples allow the detection of more than 240 metabolites related to inborn errors. This gives parents a higher chance to find possible, known or unknown inborn errors of their baby and therefore increasing the chance of an appropriate treatment, as early as possible. This can strongly increase their quality of life, giving a more secure and safe feeling to the family.

All investigations of not conspicuous newborns are helping to establish this procedure as general screening method for every newborn. So that all parents can benefit from the better screening outcome and increased quality of life. This service is offered at a special prize of only 150 € per newborn.

Analyses

Proton-containing compounds in body fluids

Description

Assay, Quantification and Interpretation

Method

$^1$H-NMR Spectroscopy

Sample

Urine, heparinized blood plasma, cerebrospinal fluid (CSF)

Costs

150 € per sample. For this we will send an invoice with payment instructions.

Turn-around time

10 days

Shipment instructions

Please download all documents from: www.infai.de → NMR → Metabo Test

Delivery Address

INFAI GmbH, Gottfried-Hagen-Str. 60-62, 51105 Köln, Germany

Contact

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References

- S. Aygen et al. “NMR-Based Screening for Inborn Errors of Metabolism: Initial Results from a Study on Turkish Neonates” (Publication)
- Metabo Test INFAI – “NMR based screening for inborn errors of metabolism” (Brochure)
- M. Spraul, S. Aygen – Urine Based Newborn Screening Project applying High Resolution NMR-Spectroscopy (Flyer)