

Questions Answered by Professor Khosrow Adeli PhD, FCACB, DABCC

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Answers are shown in RED:

1. Which methodology is used to measure urine pterins and DHPR at Sick Kids?

We send these tests to another hospital. We do not perform them in our lab.

2. What are the minimum biochemical tests we can include to screen IEM? (thinking about cost in developing country)

The minimum is two tests: Phenylalanine for PKU and TSH for Hypothyroidism

3. Is it always necessary to do genetics after definitive biochemical findings? As part of profiling for local genetic variants

Genetic testing is not essential. But it can be helpful if available.

4. If we are starting IEM screening programme in a hospital of India, which of the methods is the most cost-effective way to do so?

The best approach is to use Mass Spectrometry to screen for several inborn errors. If this is not possible the absolute minimum is two tests: Phenylalanine for PKU and TSH for Hypothyroidism

5. Is there any possibility to separate Isomeric amino acids on LCMSMS?

There is no easy way to separate them. Different columns are to be used.

6. What is the best technique for neurotransmitter analysis, and which matrix is most significant for diagnosis?

One of the best methods is UHPLC–MS/MS.

7. Can we use cord blood for screening?

Heel prick samples are preferred for newborn screening; cord blood is not recommended for screening by MSMS.

8. Can we screen metabolic diseases of newborn before the birth (during pregnancy)?

Prenatal screening can be used to screen a child for chromosomal abnormalities such as Down syndrome, trisomy 18 and open neural tube defects. But this does not replace the newborn screening that needs to be done after child birth.

9. Why GS -MS is a good method for uric acid analysis

Because organic acids are easy to vaporize. So, GC-MS is the best way.

10. I am Chemical Pathologist from Pakistan. I am highly interested in training opportunities in Canada so that we can implement diagnostic facility in our country regarding IEM and Newborn Screening.

I suggest contacting the newborn screening program in Ottawa. We do not have such opportunities in Toronto. <https://www.newbornscreening.on.ca>

11. Are there any programme or training options in the field of biochemical genetics? IFCC should take an initiative for interested candidates in biochemical genetics programs.

No international programme exists but there are biochemical genetics training programmes in the US, Canada, Germany and other countries. I agree that such training programmes should be encouraged and supported by IFCC.

12. Do you have any data regarding incidence of IEM in developing countries?

No, unfortunately such data are not readily available.

13. Is there available single reagent that can be used to analyse the panel of Inborn error in children?

No, there is no such reagent; but tandem mass spectrometry can be used to screen for multiple diseases using the same technology.