The Restoration Act (INGO) is proudly offering virtual seminars on various clinical topics of inborn errors of metabolism, genetics, and neurology in collaboration with the Ministry of Health Kurdistan Regional Government, Directorate General of Health Duhok, Iraqi & Kurdistan Boards for Medical Specialization/Pediatric, University of Duhok College of Pharmacy and Pediatric Department of Medicine, Kurdistan Pediatric Society, Hevi Hospital, and the Iraqi Pediatric Society.

Questions & Answers from:

"A Specialized Case-Based Review in Newborn Screening of Inborn Errors of Metabolism – An Approach to Diagnosis and Management"

Held 26 September 2024

Watch the recorded Virtual Clinical Seminar

Passcode: **R3fJ+4Vf**

Presented by Metabolic Specialist:

Dr. Joshua Baker, DO, FAAP, FACMG

Dr. Baker written answers to the questions as written during the live seminar:

Question: up to my experience with PKU patients, that I see that physiotherapy (exercises) can help those patients by activating the muscles of the to use the energy and absorb the metabolism and then the protein that we think on the brain of the patient it absorbed by storage system of the body or muscles to use and slowly slowly these protein will be absorbed , here my question it that only by these process we cane absorbed these protein?

Answer: PT is very helpful for many of these disorders because it forces the body to use the energy and in case of PKU lets the muscle take the high amino acids and build muscle. Very great way to treat many conditions.

Question: What are the general clinical signs or red flags that should prompt a clinician to consider an inborn error of metabolism in a patient, especially in cases where the symptoms are nonspecific or overlap with more common conditions?

Answer: Think of IEM of you see multiple systems involved or a progressive change

overtime. There is many overlap for mild disease so sometimes great to consider if the patient is not responding to treatment.

Question: It was mentioned that we shouldn't take a blood sample for screening too early, but also not too late. What is the perfect time to take the sample?

Answer: About 48 hours. However, later is better than never for some of these conditions.

Question: thanks for the fruitfull presentation regarding MSUD some regarded as aminoacidopathy and others concider it as OA so is any explenation for this conflict

Answer: Considered both by different institutions. It is the middle of being both. Hard to separate perfectly into boxes sometimes. https://www.ncbi.nlm.nih.gov/books/NBK557773/#:~:text=Maple%20Syrup%20Urine%20Disease%20(MSUD,and%20potentially%20irreversible%20developmental%20effects.

Question: How does early detection of inborn errors of metabolism through newborn screening impact the long-term neurodevelopmental and physical outcomes in affected individuals compared to those diagnosed later in life?

Answer: Most patients do better in terms of neurodevelopment with early treatment and diagnosis. Not always true for severe cases, but overall the earlier the detection and treatment, the better the outcomes.

Question: Thank you dr Joshua, excellent presentation. what is your advice to prevent this kind of disease?. A means what is the main point to decrease the incidence of metabolid disease?.

Answer: I am not sure what we can prevent. Early diagnosis and management is better for outcomes. Likely seeing more due to better testing for these disorders. Also important to education populations at high risk like those from related marriages.

Question: should blood sample for newborn screening be obtained after ensuring good feeding ? better than specific age ?

Answer: Blood samples are best after feeding, which is why 24-48 hours is best. However, sometimes the reason the baby is not feeding well is due to a metabolic disorder. If the patient is on IVF, better to be off fluid and on feeds before testing.

Question: What he know about ravicti vial

Answer: Sorry I do not understand the question.

Question: When to expect epilepsy cause as IEM not CNS disease?

Answer: If there are multiple systems involved or a progressive change overtime. Also consider if patient is not responding to treatment.

Question: Best treatment for patients with seizure due to IEM?

Answer: Really depends on the type of seizures. Often best to avoid Valproic Acid due to liver injury.

Question: Why patients either IEM have recurrent RTI?

Answer: This is two part. 1 – due to low protein diet affecting ability to make proper antibodies. 2 – some IEM have direct toxins that affect the bone marrow and/or immune system.

Question: MSUD: what will be result of blood ketone? While urine ketone is positive?

Answer: Depends on the specific ketone. Ketones in the urine are better because it is helping detect organic acids not just ketones.

Question: In FAOD; why your patient have positive detone in urine?

Answer: Apologies. That is a typo while making the presentation. VLCAD should have non-ketotic hypoglycemia.

Question: What are the cause of hyponatremia in MSUD?

Answer: It is complex, but a simplied view is there is disruption of the Na/K channels. Also water likes to move where the leucine is, so, the more leucine in the blood or brain then water is pulled into those areas causing the complications we see.

Question: Do you recommend to use vitamin B complex as blind line of therapy in suspected cases of IEM?

Answer: Vitamin B complex is very safe so always great to give as a trial while you are working up an IEM