argininosuccinate 1014 $\mu\text{mol/L}$, consistent with a diagnosis of argininosuccinic aciduria, subsequently confirmed on molecular genetic testing. She was discharged on oral sodium phenylbutyrate, sodium benzoate and L-arginine, with an appropriate dietary plan. To date, there is no evidence of neurological sequelae, likely attributable to prompt diagnosis and treatment.

Conclusion Argininosuccinic aciduria is a rare, autosomal recessive disorder of urea cycle metabolism caused by deficiency of argininosuccinic lyase. Clinical and biochemical manifestations depend on the severity of enzyme deficiency. In the neonatal variant, it typically presents with hyperammonaemia, encephalopathy and respiratory alkalosis. This case highlights the importance of considering inborn errors of metabolism in the assessment of a sick neonate.

P432

ENCEPHALOPATHY IN TEENAGERS – A CHALLENGING ETIOLOGIC DIAGNOSIS

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Introduction Inherited metabolic disorders due to intoxication provoked by highly toxic endogenous compounds are among the causes of encephalopathy. That is the case of urea cycle disorders in which ammonia's great neurotoxicity can frequently be lethal. Ornithine Transcarbamylase (OTC) Deficiency is an X-link genetic disorder and the most common disorder of the urea cycle. Presentation in heterozygote females is variable and can go from being asymptomatic to having mild chronic symptoms with severe exacerbations in any age.

Case report Fifteen year old female with sporadic headache since the age twelve without other relevant medical history. Due to persistent frontoparietal headache for two weeks, she was given topiramate. Despite medication, the headache worsened and dizziness, blurred vision, dysarthria, aphasia, asterix, motor incoordination and lethargy developed. Trauma, recent infections or toxic ingestions were denied. Diagnostic workup in a private hospital with laboratory studies including urine toxic screening, complete blood count, hepatic and renal function and cerebral MRI were normal. On account of a suspected iatrogenic cause for encephalopathy, topiramate was suspended after two days and the patient was referred to a Pediatric Emergency Department. On admission CSF study was normal but blood tests showed hyperammonemia (229 $\mu\text{mol/L}$) with latter increase to (359 $\mu\text{mol/L}$) despite treating measures (endovenous fluids and discontinuation of protein intake); She was given sodium benzoate, sodium phenylbutyrate and arginine resulting in blood levels of ammonia returning to normal. The demonstration of elevated urinary orotic acid led to the probable diagnosis of OTC deficiency which was later confirmed by molecular diagnosis.

Discussion Serum ammonia levels could be measured in every patient with unexplained altered mental status. Primary (urea cycle disorders) and secondary hyperammonemia alike are considered emergency situations therefore, treatment should never be delayed by diagnostic tests. On the other hand, both treatment and exacerbation preventions have an impact in prognosis regardless of age.

P433

AN INTERESTING CASE DIAGNOSED AS BOTH PHENYLKETONURIA AND MATERNAL PHENYLKETONURIA

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Introduction Phenylketonuria (PKU) is the most common and autosomal recessively inherited metabolic disease due to the deficiency of phenylalanine hydroxylase (PAH). Elevated levels of phenylalanine are not only toxic for the children but also teratogenic for the fetus. Clinical findings of maternal phenylketonuria (MPKU) are intrauterine growth retardation, microcephaly, significant developmental delay, congenital cardiac anomalies, and some other structural defects. Poorly diet control and high levels of blood phenylalanine causes to this severe but preventable clinical syndrome.

Case report A seven-day-old girl admitted to the pediatric metabolism clinic with a suspicion of PKU after newborn screening program. She was born at 40 weeks of gestation with a 2300 gr birth weight and hospitalized for 2 days. Her parents were first degree cousins and the family history was unremarkable for an inherited metabolic disease, however the mother had two miscarriages. Physical examination revealed microcephaly, mild facial dysmorphism, and cardiac murmur in addition to intrauterine growth retardation. Her blood phenylalanine level was 1140 $\mu\text{mol/dL}$ and she was diagnosed as moderate phenylketonuria. As a ventricular septal defect was detected with echocardiography and the mother was born before the national newborn screening program was available, the mother's blood Phe concentration was measured. Surprisingly her blood Phe level was 1614 $\mu\text{mol/dL}$. The mother was a 25 years old woman be able to graduate from primary school and complained about only concentration problems and forgetfulness. She had also blond hair and blue eyes on physical examination.

Conclusion Maternal phenylketonuria is a preventable public health problem which causes undesirable results like mental-motor retardation and cardiac defects. Although maternal phenylketonuria is not completely coped with, after newborn screening program the incidence is decreased. A strict Phe restricted diet beginning before the pregnancy together with frequent controls of blood Phe levels are essential for management. Here, we report this case, as she was diagnosed both phenylketonuria and maternal phenylketonuria and in order to emphasize thinking MPKU in the presence of microcephaly, developmental delay, and cardiac defects.

Maternal phenylketonuria, newborn screening program, developmental delay.

P434 TYROSINEMIA TYPE 1: A CASE REPORT

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Introduction Tyrosinemia type 1 is an AR inherited metabolic disorder attributed to deficiency of Fumarylacetoacetatehydrolase, which is a terminal enzyme in the metabolism of tyrosine. The gene for this enzyme has been mapped to the long arm of chromosome 15. Its prevalence has been reported as 1: 100.000. The patients with tyrosinemia expire in the early years of their lives. There is a markedly increased risk of hepatocellular carcinoma among the survivors.

Case report An Saudi 8-month-old male infant presented with complaints of abdominal distension, fever, jaundice, melena and disturbed level of consciousness for 3 days prior to admission to PICU (in King Fahd Hospital, Al-Baha). He was the 1st child of 1st degree consanguineous parents.

His motor and mental development were delayed. One day after admission, the infant developed repeated daily attacks of neurological crises in bouts of irritability, crying with increased tone and deep tendon reflexes in the lower limbs.

On physical examination patient looked sick, pale, jaundiced and drowsy. He had rachitic signs. There was mild lower limb edema. Fine crackles were audible bilaterally on the chest. Abdomen was distended. Liver was palpable 5 cm below right costal margin. Spleen was just palpable below left costal margin. There was positive shifting dullness, with scrotal edema. The infant was drowsy, with poor vision, and hypertonic, with exaggerated deep tendon reflexes.

Hematological screening CBC showed anemia and thrombocytopenia. CRP was positive and urine analysis showed albumin & RBCs.

Biochemical examination showed that BUN: 12.5 mmol/L, creatinine 27 µmol/L, glucose: 2.9 mmol/L, Ca: 2.2 mmol/L, phosphorus: 0.64 mmol/L, Mg: 0.57 mmol/L, albumin: 28 µmol/L, T. bilirubin: 35 µmol/L, direct bilirubin: 17 µmol/L, alkaline phosphatase 426U/L, AST: 65U/L, gamma GT: 130U/L, LDH: 566U/L, ammonia: 121 µmmol/L. Coagulation screening was abnormal. Blood Alpha fetoprotein was very high, ABG examination showed compensated metabolic acidosis.

Tracheal aspirate C/S showed MRSA The left wrist graphy showed evidence of rickets. Abdomen ultrasonography revealed hepatosplenomegaly with moderate ascites. Liver examination showed multiple hyperechoic masses. Abdominal CT showed hepatomegaly with multiple hyperdense masses, splenomegaly and ascites. CT brain showed cerebral atrophy.

Blood phenylalanine, tyrosine and methionine levels were all high. Urine examination showed increased levels of organic acids.

So, the infant was diagnosed as tyrosinemia type I and was prescribed a phenylalanine and tyrosine restricted diet (special formula milk) and received treatment for Vit. D resistant rickets. Following treatment, patient showed much clinical and laboratory improvement, but not to the safe level to conduct liver biopsy.

P435 RETINOPATHY OF PREMATURITY PREVALENCE AND RISK FACTORS IN DEVELOPING COUNTRIES

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Retinopathy of prematurity prevalence and risk factors in developing countries

Introduction Retinopathy of prematurity (ROP) is an avoidable disorder in preterm infants, which can lead to blindness unless it is diagnosed and treated at early stages.

Objectives The goal of this study is to determine the prevalence of ROP and its predisposing factors in preterm infants admitted to NICU at community hospital, Mashhad, Iran.

Method ROP screening was done for infants that admitted to NICU weighing less than 1500 mg and a gestational age of 32 weeks or less was conducted from January 2018 to 2019 in. A total of 149 neonates (89 males and 60 females) underwent retinal evaluation using indirect ophthalmoscopy during the fourth week and were periodically followed up.

Prenatal risk factors of ROP were evaluated using univariate analysis.

Results Of the 149 neonates under study, 42 were ROP positive out of whom 17 (40%) were at stage 2, 12 (29%) at stage 1, 2 (5%) at stage 3 and 2 (5%) at stage 4 while 19 (21%) of newborns were treated with Avastin injection. The results of univariate analysis showed that there was a significant difference between ROP occurrence and gestational age, birth weight and oxygen therapy duration. ($p = 0.0001$). However, there was no significant relationship between the ROP occurrence and gender ($p = 0.55$), stage of disease and gestation age ($p = 0.208$) and oxygen therapy duration ($p = 0.314$).

Conclusion The estimated prevalence of ROP in this study was 28% that was higher than developed countries. Gestational age, oxygen therapy and low birth weight are known risk factors of ROP. The risk of ROP in a gestational age of less than 32 weeks is 4.2 times higher than the gestational age of more than 32 weeks. Therefore, it is essential to make plan for the most effective control over the predisposing factors like oxygen therapy, preventing premature delivering, and incidence of birth asphyxia in NICU.

Retinopathy, preterm infant, prevalence, developing countries

P436 KNOWLEDGE AND PRACTICE OF PULSE OXIMETRY SCREENING FOR CRITICAL CONGENITAL HEART DEFECT IN NEWBORN INFANTS BY PAEDIATRICIANS IN BENIN

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Introduction Pulse oximetry screening for critical congenital heart defect in newborn infants is not widely used even in developed countries. In Benin, the diagnosis of congenital heart disease is often late, making the surgical treatment impossible