Study design: This prospective study was conducted on 50 pre-term neonates with RDS and 25 without RDS. On D3, sCysC, sCr, and blood urea nitrogen (BUN) were measured and estimated GFR (eGFR) was calculated. Neonates were evaluated for development of AKI during the first week of life according to the modified pediatric RIFLE (pRIFLE) criteria.

Results: Thirteen neonates with RDS developed AKI (26%). There was no significant difference between RDS group & controls regarding sCysC. RDS neonates with AKI had significantly higher sCysC than those without AKI (1.62 ± 0.12 versus 1.16 ± 0.09 mg/L; p < 0.001). Neonate with AKI grade I (failure) had significantly higher sCysC than those with R (risk) and I (injury) (P = 0.028). RDS grade III-IV neonates had significantly higher sCysC than RDS grade I–II. There was no correlation between D3 sCysC and gestational age (r = -0.454, P = -0.454) and birth weight (r = -0.442, P = 0.130). Receiver operating characteristics (ROC) curve showed that D3 sCysC can predict AKI in preterm neonates with RDS at a cutoff point of >1.3 mg/L with a sensitivity of 92.3% and specificity of 96% while D3 sCr has poor sensitivity and specificity (76.90% and 68.0% respectively).

Conclusion: Preterm neonates with RDS are at increased risk of AKI. sCysC on day 3 of life can predict AKI earlier than serum creatinine and eGFR.

Phenotypic characteristics of a tyrosinemia type I: an Egyptian cohort

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Introduction: Tyrosinemia type I (HT1) is an autosomal recessive inborn error of metabolism. The actual prevalence in Egypt is unknown but expected to be high with our 35-50% consanguineous marriages. The disease’s early manifestation can be easily missed in a country where viral hepatitis is the main physician’s concern. Current management guidelines include lifelong specialized diet and use of the orphan drug, nitisinone which is considered unaffordable to almost all Egyptian patients. Aim of work The aim of this study is to highlight the phenotypic characteristics of a cohort of Egyptian patients with HT1 including the available treatment options and outcomes.

Patients and Methods: This retrospective study included data from 21 patients’ files with HT1 who were followed by at least one of the authors. Full history taking, thorough clinical examination and investigations including succinylacetone (SA) in dried blood spot/ urine, tyrosine level, CBC, liver and renal functions, urine analysis, abdominal ultrasound and doppler and Echocardiography, slit lamp examination were done to all patients. Liver biopsy, fibroscan and multislice CT abdomen were done to selected patients. Data of different treatment modalities including liver transplantation and nitisinone were also included.

Results: The 21 patients were from 20 different families, with 80% consanguineous marriages. Family history of similar affected patients was present in 11 patients. HT1 was confirmed by SA in urine or DBS in all patients. The youngest age of presentation was 2 months and the oldest was 8 years. Abdominal distension and hepatomegaly was the main presenting symptoms (95%) followed by rickets in 11 patients (52%). Investigations revealed increased INR in 90% of patients while cholestasis and increased liver function tests in only 38%. Hepatic focal lesions were present in 52% while portal hypertensio was present in 28% of patients at presentation and abnormal echocardiographic findings in 24%. Beside the liver and kidney supportive management, protein restricted diet, 3 patients did successful liver transplantation while 11 received interrupted nitisinone capsules. Three patients died during the ten years follow up (two with HCC and one during neurological crises).

Conclusions: Hepatomegaly associated with coagulopathy (and not cholestasis) is one of the earliest signs of HT1 especially if associated with rickets which should not be missed in even in a country who is genetically susceptible to vitamin D deficiency. The recommended medical management of HT1 is a considerable financial burden to the Egyptian families and could have major side effects if interrupted and so liver transplantation should be offered to each patient.

Central adiposity and left ventricular mass in obese children

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Background: The prevalence of children obesity is increasing at an alarming rate. Most of metabolic and cardiovascular morbidities characteristic of adult obesity are likely to originate in childhood; therefore, early identification of these alterations is warranted. There is strong evidence that left ventricular (LV) mass is high in normotensive obese adults, and that it is closely associated with body mass index (BMI) and insulin resistance (IR). Aim of our study is to assess relation between central obesity and cardiac mass in obese children. Subject and Methods: The study population included 56 obese and 36 non-obese children as a control. All patients had BMI/ BMI SDS calculation, waist/hip ratio and waist/hip ratio SDS, blood pressure measurement, echocardiography for measurement of cardiac mass.

Results: Obese children showed higher BMI, Waist/hip ratio and HOMA-IR. Obese group exhibited higher levels than control group of total cholesterol, LDL and triglycerides, while a lower value of HDL cholesterol. RV MI (right ventricular mass index), LV MI (left ventricular mass index), LAD (left atrial diameter), SWT (septal wall thickness) all are higher in obese children comparing to non-obese one. Among obese subjects strong correlation between BMI SDS and PWT was found.

Conclusion: Cardiac mass is higher in obese in children in comparison to non obese children.

Ischemia modified albumin in Egyptian patients with β-thalassemia major: relation to cardiac complications

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Background: β-thalassemia is a hereditary chronic hemolytic anemia that requires lifelong regular transfusion therapy which