Prolidase deficiency

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Prolidase deficiency is a very rare hereditary disorder of collagen metabolism that leads to skin fragility and recurrent ulceration. Prolidase enzyme is widely distributed among organs and tissues, and involved in several functions, such as protein catabolism, especially collagen turnover, and regulation of collagen biosynthesis. It is also involved in cell growth, wound healing, inflammation, angiogenesis, proliferation and carcinogenesis. Patients with prolidase deficiency have accumulation of undigested dipeptides that leads to a wide spectrum of manifestation including chronic, slowly healing ulcerations, haptosplenomegaly, anemia and dysmorphic features. Here we report a family with two children affected with this disease and variable phenotypic features.

Vitamin K status in Children with Chronic Liver Disease: Do we need a more sensitive marker?

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Background and Aim: Vitamin k plays an important role in the synthesis of clotting factors, its deficiency is associated with increased bleeding tendency. Assessment of Vitamin k status is usually done through measurement of INR. The aim of this study was to compare the conventionally used INR to PIVKA II concentration in children with chronic liver disease.

Subjects and Methods: Fifty children with CLD were included in this study, they were divided to cholestatic, and non-cholestatic groups. All were subjected to clinical evaluation and lab studies including INR and PIVKA II concentration using ELISA before and after IV Vitamin K to all patients for 3 days.

Results: PIVKA II levels were elevated in all patients with CLD (60.3 ± 25.6 ng/ml). It was significantly higher among children with cholestatic CLD (81.76 ± 16.33 ng/ml) compared to the non-cholestatic group (38.84 ± 10.59 ng/ml) (p < 0.0001). All patients with bleeding tendency (17/50) had elevated PIVKA II levels, while 11 out of them (11/17) showed elevation in their INR. After IV vitamin K, PIVKA II levels showed evident reduction in both groups being more significant in the non-cholestatic group.

Conclusion: Elevated PIVKA-II levels was found in all children with CLD despite oral vitamin k supplementation and even in the presence of normal INR indicating that PIVKA-II could be a more sensitive marker of subclinical vitamin K deficiency and that Vitamin K doses, rout of administration and monitoring need to be carefully adjusted in patients with CLD.

Vitamin D deficiency in obese children and adolescents and relation to glucose homeostasis

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Background: Obesity is the primary risk factor for development of impaired glucose tolerance, and metabolic syndrome. Obese children and adolescents are at an increased risk of vitamin D deficiency because it is sequestrated by excess adipose tissue, thus lowering its bioavailability. Vitamin D is critical in both glucose homeostasis and insulin secretion.

Objective: to compare the prevalence of vitamin D deficiency in obese and lean children and adolescents and to evaluate the relation between 25 (OH) D level and markers of abnormal glucose metabolism.

Methods: This cross-sectional controlled study included 40 obese children and adolescents (age, 3- 14 years; BMI > 95th percentile for age) randomly recruited from the Obesity Clinic, Children's Hospital, Ain- Shams University and 40 age-, sex-, ethnicity- and season- matched healthy lean subjects served as controls. Clinical evaluation including blood pressure and different anthropometric measurements of patients and controls including weight, height, waist and hip circumferences and waist/hip ratio. Fasting serum glucose and insulin, fasting lipids and 25-OHD were measured. The homeostatic model assessment of insulin resistance (HOMA-IR) was calculated in both groups.

Results: The obese subjects had higher fasting glucose and significantly higher fasting insulin (p- value= 0.000), also they had significantly higher HOMA-IR (p-value= 0.000), higher LDL- cholesterol levels (p = 0.000), triglycerides and significantly lower HDL- cholesterol levels than controls (p = 0.000). The levels of 25-OHD in the obese group (16.74 ± 16.33 ng/dl) were significantly lower than those of the controls (77.44 ± 44.4 ng/dl) (p = 0.000). A significant negative correlation was found between 25(OH)D and HOMA-IR in obese subjects (r = -0.192, p = 0.000).

Conclusion: Vitamin D deficiency is common in Egyptian children and is significantly more prevalent in obese children.

Arthrogryposis-renal dysfunction-cholestatic (ARC) syndrome: a rare cause of cholestasis

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Arthrogryposis-renal dysfunction-cholestatic (ARC) syndrome is a rare autosomal recessive disorder with classical features of congenital joint contractures, renal tubular dysfunction and cholestatic jaundice. Some patients have additional features including ichthyosis, platelet anomalies, agenesis of the corpus callosum, congenital cardiovascular anomalies, deafness, recurrent infection, and internal bleeding. Here we report two Egyptian patients from two different families with this rare disorder.

Subjects and Methods: In this study, they were divided to cholestatic, and non-cholestatic groups. All were subjected to clinical evaluation and lab studies. The homeostatic model assessment of insulin resistance (HOMA-IR) was calculated in both groups. They had significantly higher HOMA-IR (p-value 0.000), triglycerides and significantly lower HDL- cholesterol levels than controls (p = 0.000). The levels of 25-OHD in the obese group (16.74 ± 16.33 ng/dl) were significantly lower than those of the controls (77.44 ± 44.4 ng/dl) (p = 0.000). A significant negative correlation was found between 25(OH)D and HOMA-IR in obese subjects (r = -0.192, p = 0.000).

Conclusion: Elevated PIVKA-II levels was found in all children with CLD despite oral vitamin k supplementation and even in the presence of normal INR indicating that PIVKA-II could be a more sensitive marker of subclinical vitamin K deficiency and that Vitamin K doses, rout of administration and monitoring need to be carefully adjusted in patients with CLD.