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Clinical case reports: Multiple pediatric case presentations - Asma Awadalla - Ministry of National Guard Health Affairs

Asma Awadalla

Ministry of National Guard Health Affairs, Saudi Arabia

From King Abdullah Specialized Children's Hospital, Riyadh, Saudi Arabia, I would like to share with other paediatricians 5 cases from the middle east: Arthrogryposis-Renal dysfunction-Cholestasis (ARC) Syndrome: Four cases from different ethnic groups but all from middle east with ARC syndrome a multisystem disorder which was reported as a rare autosomal recessive disorders. Clinical presentations, physical examination and and investigations including genetic tests were described. In three patients we also highlighted the course of the disease and age at death for this life limiting condition. A rare presentation of a complicated case of hypercalcemia: A previously healthy 11 years old boy, who presented with right flank pain for one week.

Associated with vomiting. Apart from right renal angle tenderness system exam was normal. Investigations revealed increased serum and urinary calcium with elevated parathyroid hormone level. Rt ureteric stone was evident on plain film and renal USS. The patient had laser fragmentation of right ureteric stone then partial parathyroidectomy. Histopathology confirmed parathyroid adenoma which was suspected on the parathyroid scan. Outpatient follows up reassuring. A case of Agammaglobulinemia: Nearly 4 years old boy presented with sudden onset of Rt sided weakness and left-sided facial asymmetry. He had 2 previous admissions for recurrent infections.

Neuroimaging consistent with occlusion of left MCA and sub-acute infarction involving fronto-pareital lobes. Echo shoed depressed cardiac function and immune work up showed Zero Bcells consistent with Agammaglobulinemia. Stroke was likely thrombo-embolic secondary to cardiomyopathy/ LV dysfunction as he had dilated left atrium and left ventricle with mild to moderate mitral insufficiency and moderately depressed left ventricular systolic function.

Patient was started on regular IVIG. Two cases of disseminated BCGgitis secondary to interleukin 12 deficiency and BCG-osis: The first case is an 18 moth old boy presented with dissaminated BCG infection (left axillary lymph node and splenic abcecces) secondary to IL-12 deficiency on antiTB medication. The second case is a six month male infant with BCG related axillary lymphadenitis.

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