SELECTED ABSTRACTS FROM NATIONAL MEDICAL JOURNALS

Pages with reference to book, From 239 To 239

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CONGENITAL ERYTHROPOIETIC PORPHYRIA. A CASE REPORT. Khan, I., Hakim, I., Khan, F.M. Pak. Paed.J., 1990; 14:179-182.

The Case of a 6 years old boy diagnosed as congenital. erythropoietic porphyria is presented. The presenting complaints were erythcma and blistering over the face and hands, red coloured urine and reddish brown discolouration of teeth. The symptoms were noted since 8 months age. The skin lesions developed more on exposure to sunlight. The vesicles would rupture leading to ulceration, crusting and healing with scarring.

The child was severely anaemic. Bullae were present on the face and hands where pigmented and depigmented areas of the skin were also noted. Hypertrichosis was absent and teeth were discoloured reddish brown. Nails were dystrophic and discoloured. Moderate splenomegaly was present. The rest of the systemic examination revealed no abnormality.

The blood picture revealed signs of haemolytic anaemia with a Hb of 6.80% and reticulocytes 7.5% with polychromasia, microcytosis and anisocytosis. LFTs revealed increase in unconjugated bilirubin. Blood porphyrin could not be estimated due to lack of facilities. Porphyrin in urine was positive.

The child's parents were first cousins and a one year old sister had pink urine and red coloured teeth. Classical genetic, clinical and laboratory features lead to the diagnosis of CEP in the child.

Congenital erythropoietic porphyria also called Gunther's disease is a rare autosomal recessive disorder characterised by severs cutaneous photosensitivity, red urine and haemolytic anaemia. The manifestations appear in infancy and the primary abnormality is a decreased activity of uroporphyrinogen accumulation and hyperexcretion of type I uroporphyrins and corporphyrins.

Avoiding sunlight, prevention of trauma and treating secondary infection are the mainstayof management. Packed red cell transfusion, splenectomy and oral charcoal have been tried as therapy to decrease porphyrin excretion and decrease haemolysis.

CONN'S SYNDROME. Salahuddim, N. Pak. J. Surg., 1989; 5: 15-17.

The case of a 30 years old female diagnosed as Aldosteronism is presented. The patient presented with poorlycontrolled hypertension of two years duration. The past history was insignificant and the drug history gave an irregular intake of propranolol, prazosin, labetalol or thiazides.

The woman came in with a headache and a blood pressure of 200/120 mmHg, supine in both arms with no significant postural drop. The systemic examination and fundi were normal. The laboratoty tests showed alow serum potassium of 2.4 meq/dl. The other routine tests were within normal limits.Arepeated serum K after oral supplementation for a weekwas still low; 3 meq/dl. A 24 hour urine VMAwas normal.

The persistent hypertension and hypokalaemia provided a suspicion for hyperaldosteronism. A CT scan of the abdomen revealed a right sided adrenal mass which was confirmed on renal arteriogram. There was no renal artery stenosis.

Exploratory laparotomy showed the right adrenal cortical adenoma which was resected. Oral potassium was supplemented for 2 weeks. The patient has been normotensive and asymptomatic for 3 years after surgery.

In a young patient with hypertension secondary causes must be suspected. Hypokalaemine not responsive to potassium supplements is a strong reason to suspect hyperaldosteronism. Aldosteronism is associated with hypersecretion of aldosterone. It can be primary due to an adenoma or hyperplasia which was originally described by Conn. secondary aldosteronism is due to an extra-adrenal stimulus

and is associated with elevated plasma renin levels.
Resection of adrenal adenoma results in a complete cure of hypertension and hypokalaemia for one to five years in 70 percent of the cases and permanently in half the cases.

PENETRATING NECK INJURY. Khan, T.F.T. PalcJ. Surg., 1989; 5: 18-19.

A 5 year old boy sustained a penetrating injury by a stick in the leftside of his necknear the angle of the mandible afterafall. The child was dazed. The wound was drywith no cranial nerve deficit.
The wound was explored after establishing an intravenous line. The stick had passed through the bifurcation of the common carotid and lodged in a vertebral disc. Extraction was followed by a great spurt of arterial blood. The tear on the anteriorwall of the carotid arterywas repaired using 5% arterial silk in continuous sutures. Postoperatively, the child developed weaknesiof his right upper limb which improved in a month.
Penetrating neck in injuries can cause major vascular damage. Late exploration can lead to 65% mortality. Earlyexploration has 2-10% mortality in positive cases and 1-2% in negative ones. Emergency angiography in all penetrating injuries will help the surgeon in his decision to explore or observe. Endoscopy is required in tracheoesophageal injuries. Atemporaryshunt to preserve cerebral circulation will reduce the risk of hemiplegia. Prolene is best used for arterial repair.