SELECTED ABSTRACTS FROM NATIONAL MEDICAL JOURNALS

Pages with reference to book, From 205 To 206

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SICKLE CELL DISEASE: IS IT A RARE ENTITY IN PAKISTAN? Shamim, S., Isani, Z., Arif, MA., Ramzan, A.?

Thirteen children diagnosed as sickle cell disease are presented. A detailed history alongwith the family history was obtained. When sickle cell disease was suspected, sickling tests and hacmoglobin clectrophoresis was performed. There were 10 males and 3 females with 8 in the 2 to 5 years range, 2 in the 6-10 years group and 3 were over 10 years of age. There were 2 siblings and 2 fIrst Cousins. All the patients belonged to different ethnic groups.

The predominant symptom was repeated episodes of joint pains and progressive pallor. Sudden pallor, recurrent limp, severe abdominal pain and weakness of the right upper limb, and loss of speech were other symptoms. Severe anaemia and hepatosplenomegaly were present in 4 cases.

Painful crisis due to vaso-occiusive phenomena was a common episode. Acute splenic sequestration crisis and hyperhacmolytic crisis were observed in one child each.

The diagnosis was based on the results of haemoglobin electrophoresis. 3 cases were pure sickle cell anaemia, 4 were associated with thalassacmia and 6 had an abnormal pattern showing B-D haemoglobin.

Sickle cell disease is found in Africa, Turkey, Greece, Mediterranean countries and South Incaa. The manifestation of the disease are due to an accelerated haemolytic process or widespread occlusion of blood vessels by the distorted red cells. Episodic events or crisis threaten the comfort of life of the child. Diagnosis is made by carrying out the sickling test and haemoglobin electrophoresis. The study carried out concluded that sickle cell disease exists in Pakistan and is prevalent in all ethnic groups. Sickling test is simple and easily available for diagnosing hacmoglobin-S. llaemoglobin electrophoresis on agar gel on acid p11 differentiates Hacmoglobin S from 1).

HARLEQUIN FETUS - A CASE REPORT. Sami, A.R., Sami, A.L., Khan F.S. Pak. Ped. Journal, 1989; 13: 289-291.

Harlequin foetus is a rare skin disorder inherited as an autosomal recessive trait. The infant is usually premature and is either still born or dies in a few weeks after birth. The case of a 4051 g female infant born preterm to a 29 year old primigravida is presented. The parents of the baby were close relatives. The skin of the infant looked like the bark of the tree with reddish brown fissures dividing into triangular and polygonal shaped plaques. There was marked ectropion, gaping fish mouth deformity or eclabion, a distorted nose, rudimentary pinnac and hypoplastic pinnae. ‘Ihc hardened skin restricted respiratory movements, sucking and swallowing and caused flexion deformity of all joints. Routine laboratory tests were normal. The infant died on the seventh day after developing pneumonia.

Defect of keratinization of the skin leads to a harlequin foetus. Xray diffraction analysis of the stratum corneum in one case revealed the presence of an unusual fibrous protein suggesting a defect in epidermal keratin. The epidermal lipid metabolism is defective resulting in increase in cholesterol and triglycerides. Therapy of these cases is ineffective and most infants are either still born or expire soon after birth.

PREVALENCE OF MALNUTRITiON IN PRE-SCHOOL CHILDREN. Ahmed, S., Salahuddin , A.N.M. Pak. Ped. J., 1989; 13: 299-303.

A random prospective study was done on 200 pre-school children ages ranging between 0 and 5 years to determine the prevalence of malnutrition and to detect the important risk factors related to it.

There were 108 boys and 92 girls and each one was questioned about period of breast feeding,

immunization, socio-economic condition and family size. A physical examination was done and the body weight noted. The malnutrition was determined according to the Gomez classification.
Of the 200 children, 135 were found to be malnourished. 61 were in Grade 1, 48 Grade II and 26 were Grade III malnourished. 65 children were normal. The results indicated that a significant degree of malnutrition started at the early age of 0 to 6 months. The major risk factor was determined as lactation failure. Of the bottlcfcd babies 92.3 percent were malnourished. The other important risk factor found was delayed weaning. 82.3 percent of the children were weaned after 6 months of age in the study. The other significant risk factor was a large family size and low family income. The recommendations put forward after the study concluded were/ encouragement of breast feeding, early weaning, immunization against infectious diseases and small family size.

URBACH WIFThE DISEASE IN AFGHAN REFUGEES. Hakim, I., Khan, F.M. Pak. Pediat. J., 1990; 3: 175-178.

The case of a 9 month old female Afghan baby diagnosed as Urbach Wiethe disease is presented. This is a rare autosomal recessive systemic disorder also called Lipoid Protcinosis first reported in 1908 by Siebenmann. The etiology is unknown but it is considered to be due to primary perturbation of collagen. There is deposition of hyline like material in the dermis, sub-mucosa and other organs. Typical mucocutaneous changes are the presenting features. There is no specific treatment. Corticosteroids, lipocaic and dimethyl-suiphoxide have been tried. The disease does not affect the life span. The Afghan baby was referred due to a hoarse voice and skin lesions which she started developing since 3 months. They would start as vehicles, become crusted and then turn to atrophic scars. Eroded areas of skin and impetigo like crusts were found over elbows, knees and trunk. Hair was sparse and patches of hypopigmentation were found around the lesions. The tongue was smooth and firm but was mobile and the rest of the oral cavity mucosa showed minimal infiltration. Systemic examination revealed no abnormalities and funduscopy showed no retinal changes. Routine laboratory tests were within the normal range. The diagnosis was confirmed by a skin biopsy. Light microscopy gave the findings of mild hyperkeratosis with pink acellular hyalinised bands of callages in the upper dermis. The infant's parents were first cousins and an elder brother of 10 years age in Afghanistan had a similar illness. The overall clinical and histological features of the case were consistent with the diagnosis of Urbach-Wiethe disease.