Medical Memoranda

Congenital Ichthyosis, Spastic Diplegia, and Mental Deficiency

Sjögren and Larsson (1957) described 28 cases of the syndrome of mental deficiency, congenital ichthyosis, and spastic diplegia in Sweden. The family data indicated the mode of inheritance to be monohybrid autosomal recessive. They considered that the syndrome had not been previously reported, but three brothers described by Pisani and Cacchione (1934) are probably examples of it.

In the only family so far reported in this country (Richards, Rundle, and Wilding, 1957), two of four sibs, both girls, were affected. The parents, who were unrelated, and a brother and sister were normal. Both affected children had mental deficiency, congenital ichthyosis, and symmetrical spastic diplegia. The younger child, aged 5 years, was more mildly afflicted in all respects. Her I.Q. was 60. The family was Greek-Cypriot, and had recently migrated to this country.

The present case is of a girl of 13 years, the child of English parents who are first cousins. The parents and the younger half-brother are normal.

Photographs showing scaliness, pigmentation, and thickening of skin.

CASE REPORT

The affected girl has an intelligence quotient of approximately 40. She has spastic diplegia, but is able to stand in walking-callipers. The congenital ichthyosis is somewhat more severe than in the two girls referred to above. Scaliness, pigmentation, and thickening of the skin are particularly pronounced on the abdomen and on the neck, but are recognizable over most of the body (see photographs). She is able to smile, and speaks with difficulty. She is not epileptic and is of a placid and sociable disposition. In this, as in the other cases described, the condition appears to be stationary.

Routine urinalysis produced normal results. Chromatographic examination of urine showed normal excretion patterns of amino-acids, keto acids, and indoles. The blood picture is normal.

COMMENT

Sjögren and Larsson observed degeneration of the pigmented epithelium of the macula and its surroundings in 3 of their 28 cases. This is not apparent in any of the patients we have examined.

The Swedish authors estimated the gene frequency to be less than 0.0005 in the Swedish population. The two patients in this hospital (the third is at home) are drawn from a population of 2,200, which includes 260 patients with spastic paralysis of various types. No other patients in the hospital have congenital ichthyosis.

For comparison, there are 11 patients with phenylketonuria and 10 with tuberous sclerosis.

Rud (1927) described a syndrome characterized by congenital ichthyosis, epilepsy, dwarfism, infantilism, macrocytic anaemia, and polyneuritis. Later reports of Rud's syndrome (van Bogaert, 1935; Stewart, 1939; MacGillivray, 1954; and Ewing, 1955-6) have included mental deficiency, ichthyosis, epilepsy, signs of sexual immaturity, and sometimes other symptoms. The condition we have described appears to be genetically and clinically distinct from Rud's syndrome.

Ichthyosis uncomplicated by other symptoms is generally considered to exhibit an autosomal dominant mode of inheritance.

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Diesel-oil Injection Injuries

Diesel-oil injection injuries are not common. Published cases are few and the serious damage than can, and usually does, follow these injuries is not generally appreciated.

In diesel engines fuel is forced through fine jets at a pressure of as high as 6,000 lb./sq. in. to produce finely divided particles for rapid combustion. At such pressure