CONGENITAL ABSENCE OF PAIN

A FAMILY STUDY

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A family study is presented in which five of the 10 children have congenital absence of pain, inability to sweat and mental retardation; three children have neuropathic joints. The differential diagnosis, management and prognosis of this rare disorder are discussed.

The absence of a sense of pain as an inherited condition was first described by Dearborn in 1932. There are very scanty references to it in the orthopaedic literature; Silverman and Gilden (1959) referred to the radiological changes. Whilst it is primarily a neurological disease the manifestations which usually bring the patient to hospital are orthopaedic problems: Charcot joints, apparently spontaneous fractures, trophic ulceration and scoliosis. Despite the senior author's long experience of scoliosis and an associated orthopaedic clinical genetic unit (directed by Dr Ruth Wynne-Davies) only one such child had previously been seen.

THE FAMILY

The family recently presented at the Al-Adan Hospital, Kuwait. The parents are both originally Iraqi, both are normal but are first cousins, the commonest pattern of marriage in many Arab countries. There are 10 children, seven male and three female. Amongst these children five are normal and five affected (Table I).

The father has been able to distinguish early and with ease between the normal and the abnormal: the boys quickly demonstrate their toughness in fighting since they cannot be hurt! He has also noticed in the very hot climate of Kuwait, which averages over 120 degrees Fahrenheit (40 degrees Celsius) shade temperature for three months and may go over 130 degrees Fahrenheit (50 degrees Celsius), that none of the affected children can sweat. In hot weather he has to cool them by pouring cold water on them. Despite this, the oldest girl once had convulsions, presumably from hyperthermia. In the winter, in contrast, they feel very cold and need to be kept warm with many blankets although the temperature is never as low as zero degrees Celsius. From this it would appear that they not only lack the sense of pain but also lack temperature control. All five affected children are mentally backward, and although the older ones can read and write they spend on average three years in each class before moving up.

Table I. The children

Number	Age *(years)*	Sex	Normal	Affected
1	20	Female		*
2	18	Male	*	
3	17	Male	*	
4	14	Female		*
5	13	Male	*	(undescended testicles)
6	11	Female	*	
7	8	Male		*
8	5	Male		*
9	4	Male		*
10	3	Male	*	

On neurological examination they are normal in motor power and reflexes. Cutaneous sensation to light touch is normal, pinprick is felt but not as pain. If the pin is pushed through the skin they are aware of an uncomfortable feeling but it is not painful. The seventh child has areas over the costal margins and thighs where he can feel pain.

CASE HISTORIES

Child 1, a girl aged 20 years who has an atrophic Charcot destruction of her right hip (Fig. 1). The hip telescopes up and down several inches and has virtually unlimited, painless movement. She has thick fissured skin on the left foot with trophic ulceration. There are Charcot changes in the ankle and foot (Figs 2 and 3). The right foot was partially amputated for gangrene which developed when traction was applied for the Charcot hip. There has recently been a painless fracture of the left tibia with abundant callus. She is extremely disabled and can only walk with assistance.

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