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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REFERENCES

- Bogaert, L. van (1935). *Rev. neurol.*, **63**, 353.
Ewing, J. A. (1955-6). *Amer. J. ment. Defic.*, **60**, 98, 307, 575, 799.
MacGillivray, R. C. (1954). *Ibid.*, **59**, 67.
Pisani, D., and Cacchione, A. (1934). *Riv. sper. Freniat.*, **58**, 722.
Richards, B. W., Rundle, A., and Wilding, A. St. J. (1957). *J. ment. Defic. Res.*, **1**, 118.
Rud, Einar (1927). *Hospitalstidende*, **70**, 525.
Sjögren, T., and Larsson, T. (1957). *Acta psychiat. (Kbh.)*, Suppl., 113.
Stewart, R. M. (1939). *J. ment. Sci.*, **85**, 256.

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

the spray may easily penetrate the skin and injure subcutaneous tissues.

CASE REPORT

A man aged 42 injected a diesel fuel and paraffin (kerosene) mixture at a pressure of 120 atmospheres into the pulp of his left index finger while checking a diesel injector. He was seen in the casualty department of Queen Mary's Hospital for the East End the same day. The finger was tense and tender but it was treated conservatively with a magnesium sulphate dressing. The next day the finger was swollen, tense, and extremely painful. Lymphangitis was present on the dorsum of the hand. Penicillin was given intramuscularly, and the hand was elevated. Later the same day the terminal pulp space was incised and a through-and-through drain inserted. A relieving lateral incision was made along the medial side of the middle phalanx.

The subsequent clinical course was most unsatisfactory. The lymphangitis rapidly subsided. The proximal phalanx

was incised to relieve pain and tension on the fourth day, but the finger failed to settle. On the eighth day the patient was admitted to hospital and the finger widely opened bilaterally down to bone. The terminal phalanx was already devitalized. From then on the finger improved. Swelling and inflammation subsided and the devitalized terminal phalanx became

dry and gangrenous. A line of demarcation was present by the twenty-second day. Fig. 1 shows the finger at this stage, with gangrenous terminal phalanx and the well-healed lateral incision. The finger was amputated through the middle phalanx. The wound healed slowly and unfortunately a volar scar resulted. Subsequently, however, there was a painless, useful stump with good function.

COMMENT

Two main factors converted this apparently trivial type of injury into a serious one—the injected material and the increased interstitial pressure. Dial (1938) injected small amounts (1 mm. to 1 ml.) of fuel oil beneath the skin of a rabbit's ear and in each case a sterile abscess developed, but without the local heat and redness usually associated with infections. Thus a certain amount of tissue damage must be expected. This, combined with the increased interstitial tension caused by the injected material, caused necrosis and gangrene in practically every reported case.

Rees (1937) reported a case in which the affected finger and the distal half of its metacarpal were lost. Hughes (1941) reported a case in which two affected fingers were lost at the metacarpophalangeal joints. Dial's two patients, Golay and Verdan's (1956), and ours were more fortunate. They lost only part of the affected fingers.

The clinical course of reported cases may be divided into three phases (Golay and Verdan, 1956). (1) Immediately after injury the point or points of entry may or may not be discernible. The finger is tense and may be pale from ischaemia. Numbness of the finger gives place to pain and in the next few hours the pain may be severe enough to require morphine. (2) In the next seven to ten days the swelling increases. Pain increases

but eases as gangrene and a line of separation develops. Secondary infection may occur and spread beyond the affected finger to the palm. (3) After separation of the slough, either surgically or naturally, healing may be slow, and when attained movements of the remaining stump may be limited owing to fibrosis.

Treatment should aim at obtaining a normal finger with no loss of flesh or function. The results of expectant treatment are bad. The results of early and active treatment for the injury are unknown; but Bell (1952) and Tempest (1953) advise such treatment and report better results for the similar and better-known high-pressure grease-gun injuries.

The following is, in my opinion, the treatment of choice.

(1) The injury should be treated as a surgical emergency, and workshops where it may occur should be warned to send men suspected of having it, however apparently trivial, to hospital at once.

(2) The patient should be prepared for a general anaesthetic, A.T.S. administered, penicillin given, and as much oil as possible expressed from the puncture wound.

(3) Under general anaesthesia, the finger should be opened by full-length bilateral incisions, avoiding digital nerves and vessels but relieving tension. The tendon sheaths should be inspected and opened and irrigated with penicillin if necessary. The wound edges should be kept apart by the insertion of Penrose drains. Drainage through web-space or transverse incision in the distal palmar crease might well be added when the point of entry is the base of the finger. Dressings should be applied and the affected parts immobilized by a plaster slab in the position of function. A St. John's sling to elevate the hand is preferable.

(4) Drains should be removed on the third or fourth day, or left longer if necessary. If secondary infection can be avoided, the finger should be allowed to settle. Within ten days the exact amount of tissue damage can be assessed and then treated according to normal surgical principles of hand surgery.

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REFERENCES

- Bell, R. C. (1952). *Brit. J. plast. Surg.*, 5, 139.
Dial, D. E. (1938). *J. Amer. med. Ass.*, 110, 1747.
Golay, L., and Verdan, Cl. (1956). *Z. Unfallmed. Berufskr.*, 49, 115.
Hughes, J. E. (1941). *J. Amer. med. Ass.*, 116, 2848.
Rees, C. E. (1937). *Ibid.*, 109, 866.
Tempest, M. N. (1953). *Univ. Leeds med. J.*, 2, 125.

Domestic Food Consumption and Expenditure, 1958, the annual report of the National Food Survey Committee, contains a new feature this year—a study of the effect on the diet of the housewife's going out to work to supplement the family income. Allowing for expenditure on meals out, which is greater when the housewife is working, families with one child in which both parents were earning spent on average 4s. 6d. a head each week more on food than families of the same size dependent on one earner; for families with four or more children, the difference was 3s. a head. The diets were also more varied. But, partly owing to differences in family composition—in families with two earners the children tend to be rather older—physiological requirements were also greater, and in general the nutritional status was no higher. The report also contains a study of the diets of households which are economically dependent on a widow or single woman. Surprisingly, they fared rather better nutritionally than corresponding households in which there was also an adult male. (H.M.S.O., price 10s. net.)