bile duct perforation, or resulted from the abnormal flow of bile through the duct once the perforation had occurred, must remain a debatable point.

We report this outcome in our patient as a guide to others who may meet this condition in circumstances similar to our own. We had planned to operate, but were prevented by an overwhelming but unidentified infection which, according to physical signs and investigation, was certainly not peritonitis. At this point, our patient had already spontaneously overcome her obstructive jaundice and biliary ascites.

It therefore seems wise to treat the chronically ill patient with intravenous fluids and antibiotics as well as paracentesis for a short interval and to await spontaneous relief of the obstructive jaundice, which has been observed in most cases. The risks of surgery itself are not negligible, since Lilly *et al.*[4] mention 7 patients who died after surgery, and 1 of their 2 patients died from sepsis in the post-operative period.

We thank the Secretary of Health for permission to publish.

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Focal Dermal Hypoplasia (Goltz Syndrome)

Case Reports

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SUMMARY

Two Cape Coloured children, both with physical stigmata of Goltz syndrome, are described. Accurate diagnosis of congenital defects in the newborn allows optimal planning of surgical treatment and more accurate prognosis and genetic counselling.

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In this article, 2 patients with a well-known ectomesodermal disorder, Goltz syndrome,[1] are described, to emphasize the importance of multidisciplinary evaluation of birth defects. It is often stated that very little can be done for children with multiple congenital abnormalities. Some of these children may have normal intelligence, however, and continued supportive care, accurate treatment and prognosis can help to avoid guilt and uncertainty over future family planning, especially when the first child is abnormal. These 2 patients seem to be the third and fourth cases of Goltz syndrome reported in the South African medical literature.[2,3]

CASE REPORTS

Patient 1

This patient was a Cape Coloured baby girl whose congenital abnormalities were described as ectopia cordis and lobster-claw deformities of the hands. The chest anomaly was in fact a split sternum through which the pulsating heart could be seen. At operation the heart was found to be normal and the defect was successfully closed. The

patient was afterwards transferred to the orthopaedic department for corrective surgery of her hand deformities.

In view of her numerous physical abnormalities, the genetics service was consulted. The antenatal and delivery history as well as a family history was uninformative. The patient's mother gave no history of miscarriages and the 3 siblings from the same parents were normal.

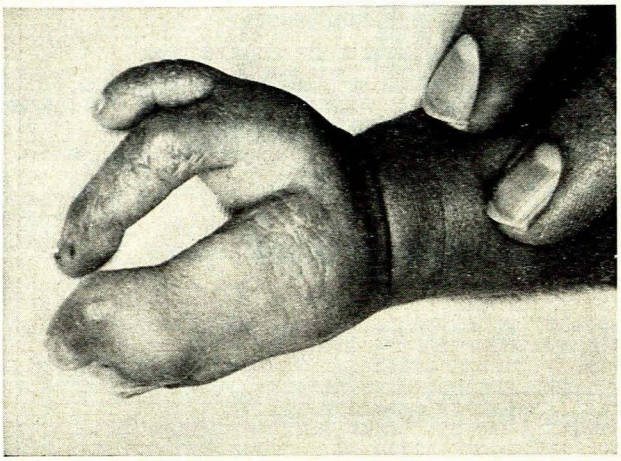

Fig. 1. Right hand of patient 1.

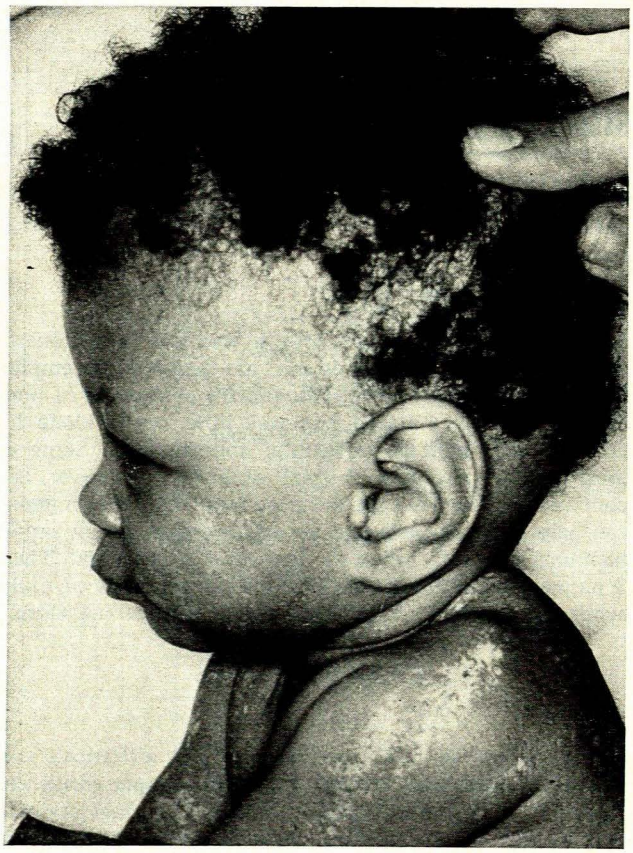

Fig. 2. Focal dermal hypoplasia over the deltoid region and patchy alopecia.

On examination at the age of 3 months the patient appeared to be of normal intelligence and developmental milestones were within normal limits for her age. The most obvious abnormalities were a split right hand (Fig. 1) with bony and cutaneous syndactyly of digits 1, 2 and 3, as well as bony and cutaneous syndactyly of the second and third toes of the right foot. The left hand had a simian palmar crease. Numerous areas of patchy hypopigmentation were seen over the body, with focal dermal hypoplasia and herniation of underlying fatty tissue.

In addition to patchy alopecia, decreased pigmentation and telangiectases were noted on the right occipitoparietal area (Figs 2 and 3). A radiological survey showed osteopathia striata in addition to the abovementioned skeletal abnormalities. These clinical findings are all typical of Goltz syndrome.[2]

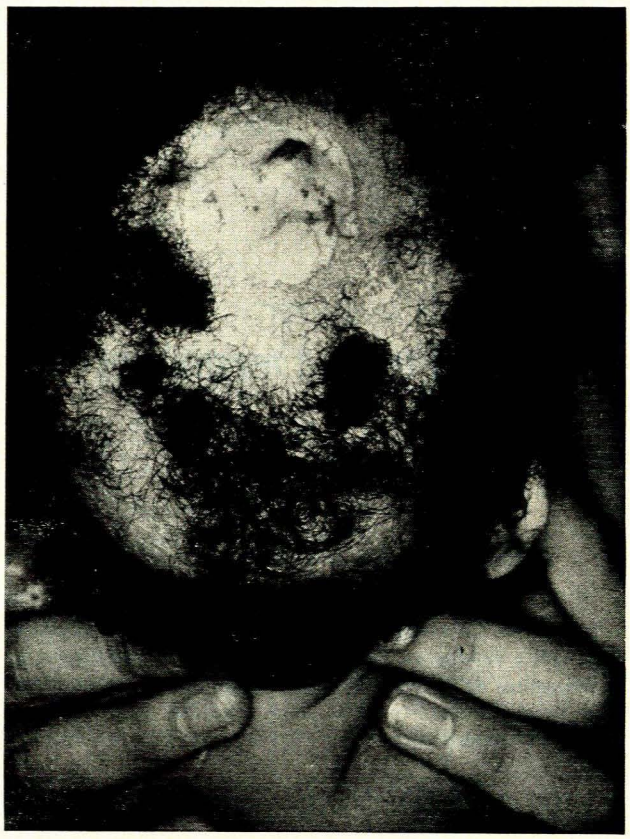

Fig. 3. Alopecia, telangiectases and fatty tissue herniation can be seen over the vertex.

Histopathological examination of the skin confirmed the focal thinning of the dermis and showed collagen arranged in thin fibres rather than the usual bundles. In some places, fat was present under the epidermis but there were no other subepithelial structures (Fig. 4). Follow-up at the age of 9 months showed normal development, and orthopaedic/plastic surgical procedures are now being planned.

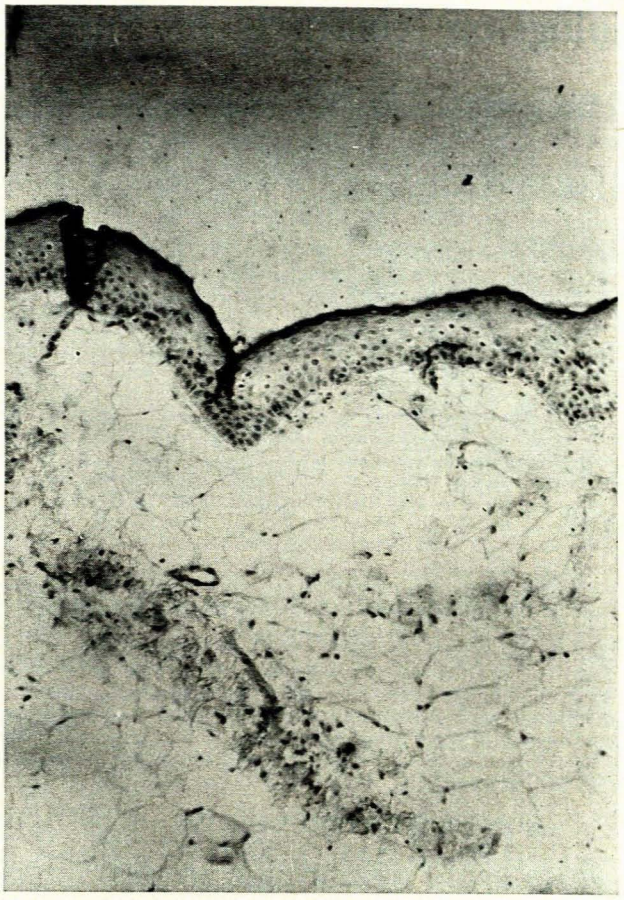

Fig. 4. Thinning of dermis with loss of subepithelial structures.

Patient 2

This Coloured patient was referred from an orthopaedic hospital for genetic evaluation. As with the first patient this was the only affected child in a family of 3 other children, and no further information was gained from the obstetric, developmental or family history.

The patient was already 2 years old and past the age at which treatment should have started to achieve the best results. The similarity to the first patient was striking. The physical examination again showed normal growth and development. Both hands had bony and cutaneous syndactyly, viz. digits 1 and 2 on the right side and 3 and 4 on the left side. The left foot displayed prominent ectrodactyly (Fig. 5) and the right foot was normal.

Patchy alopecia and hypopigmentation confined to the left knee and around the anus completed the clinical picture. Osteopathia striata was seen radiologically and it was deemed unnecessary to subject the child to a skin biopsy.

DISCUSSION

These patients represent the problem of sporadic abnormal phenotypes within large normal families. Accurate clinical

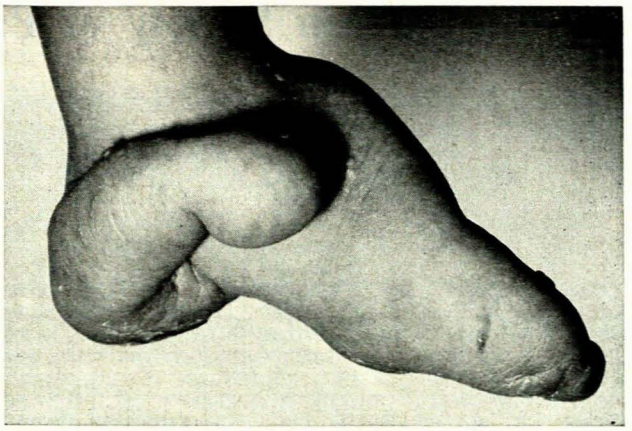

Fig. 5. Ectrodactyly of the left foot of patient 2.

diagnosis allows identification of the recurrence risk and precise prognosis associated with a characteristic anomaly or syndrome. This is the starting point for either an intensive or relatively passive approach to the problem. Goltz syndrome is an autosomal dominant condition and isolated cases in large families most probably represent new mutations. This correlates with previous reports that all cases described thus far, with the exception of 2, have been isolated examples. Some workers believe that the disorder may be due to an X-linked dominant trait, lethal in the male and with reduced fertility in the female. A few male patients have, however, been described.

The practical conclusion is that there is no recurrence risk in the families of our 2 patients and that mental retardation is a frequent but not a universal finding. With no signs of retarded development in our patients, continued follow-up and aggressive treatment seem necessary.

The aim of orthopaedic and plastic surgery is to achieve optimal functional and cosmetic results. While function and appearance are important for the hands, one would be satisfied if only function could be restored in the case of the feet.

Hand operations should ideally be done before the child starts using its hands appositionally at the end of the first year. Amputation is necessary when two fingers have bony syndactyly at their base. In the case of patient 1 (Fig. 1) adequate appositional thumb function requires sacrifice of the rest of the fused fingers. A functional foot should be attained before the baby assumes an upright position. This is done by means of centralization operations.

Long-term care for infants with malformations as outlined above, strives towards sound psychological development and minimal physical disabilities, especially when the condition is associated with a normal or near normal intelligence.

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