anuria, the treatment of choice is immediate retrograde catheterization of the ureters and irrigation of the renal pelves with warm 5% sodium bicarbonate solution. However, the mechanical effect of irrigation may be of greater importance since our patient developed a massive diuresis within a few hours of retrograde pyelography. Occasionally it has been necessary to perform nephrostomy when the ureters were completely blocked by crystals. Conservative management of the condition by administration of intravenous sodium bicarbonate without irrigation of the renal pelves will allow ultimate recovery in most patients (Arneil, 1958). However, they are likely to be subjected to the risks of unnecessarily prolonged anuria and of severe hypertension with encephalopathy.

The incidence of this serious complication of sulphonamide therapy is low, but it should be lower still. Sulphadiazine is one of the least soluble sulphonamides (18 mg/100 ml urine at pH 5.5) and without alkalinization of the urine it may cause crystalluria in 25 to 30% of patients (Weinstein, 1970). Administration of sodium bicarbonate to maintain a urine pH of 7.5 will increase the solubility of sulphadiazine to 200 mg/100 ml, but routine alkali therapy is unnecessary if a fluid intake of at least 700 ml/m² per day is given and the maximum dose of 100 mg/kg per day is not exceeded (Weinstein, 1970). Sulphadiazine has long been recommended for use in meningococcal meningitis because of the high CSF levels (60–80% of the blood level) which it achieves. However, sulphadimidine is much more soluble in urine (100 mg/100 ml at pH 5.5) and also achieves adequate CSF levels for the treatment of meningitis (Black, 1970). If a place still exists for sulphonamides in the treatment of acute bacterial meningitis in children (Wehrle, Mathies, and Leedom, 1969) then a soluble compound such as sulphadimidine should be the drug of choice.

Summary

A 3-year-old boy developed anuria due to sulphadiazine crystalluria. Bilateral ureteric catheterization and irrigation of the renal pelves was followed by the restoration of a normal urine flow rate. The use of sulphadimidine rather than sulphadiazine in the treatment of bacterial meningitis is recommended.

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Down's syndrome with diabetes mellitus and hypothyroidism

We report a girl with Down's syndrome, who has been shown to have both diabetes mellitus and hypothyroidism.

Case report

The patient was born in August 1966, a normal delivery after 38 weeks' gestation, birthweight 2.5 kg. Her mother was aged 23 years and had undergone subtotal thyroidectomy some time before pregnancy. Apart from a maternal uncle who is retarded and epileptic, there is no relevant family history. There is one normal older sister.

The child showed typical features of Down's syndrome which was confirmed by chromosome studies showing regular trisomy 21, with a total of 47 chromosomes. Her developmental progress was subsequently recorded as being slow.

In November 1967, at 15 months of age, she was admitted to this hospital in coma. She was found to have diabetic ketoacidosis with an initial blood sugar of 2000 mg/100 ml, which was successfully managed, though her diabetes later proved to be brittle and difficult to control.

In June 1972, progressive enlargement of the child's abdomen was noted and her diabetes again became unstable. She was readmitted to hospital where examination showed the presence of ascites and hepatomegaly (Fig.).

Investigations showed Hb 12.1 g/100 ml, erythrocyte sedimentation rate raised to 75 mm/hr, with the rest of the blood count normal. Liver function tests showed

FIG.—Patient at time of presentation, showing ascites.

normal serum bilirubin and no evidence of hepatocellular dysfunction or biliary obstruction. Total serum proteins normal at 8.2 g/100 ml, lipoprotein electrophoresis showed a raised β fraction, serum cholesterol was high at 345 mg/100 ml, and serum triglycerides increased at 310 mg/100 ml. These abnormally high values persisted, as did the ascites, after the child's diabetes was brought under control; and at this stage her thyroid function was investigated.

Her height at 96 cm was well below the 3rd centile, though bone age was only marginally less than chronological age. Serum protein-bound iodine was 3 μ g/100 ml (normal 4–7 μ g/100 ml) and serum thyroxine 3.6 μ g/100 ml (normal 4.5–13 μ g/100 ml). The 4-hour uptake after ^{132}I (5 μ Ci) was 13% (normal range 10–30%), but when repeated after administration of 20 units thyroid-stimulating hormone, 4-hour uptake was only 9.5%. The absence of any extra response to thyroid-stimulating hormone indicates an absence of thyroid reserve in the patient. No thyroid antibodies were shown in either the patient or her mother.

After a diagnosis of hypothyroidism, her progress on a small dose of thyroxine (0.025 mg daily) has been encouraging. She has grown 3 cm in 6 months, ascites and abdominal distension are no longer evident, and her skin and hair are less dry and coarse. Her mother reports she is generally more lively and interested.

Discussion

This case is presented not only because the combination of Down's syndrome with diabetes mellitus and hypothyroidism is rare, but also because of the unusual presentation of this child's disturbance of thyroid function.

Thyroid function in Down's syndrome is usually normal (Marks and Hamlin, 1967), but both hyperthyroidism (Kurland *et al.*, 1957) and

hypothyroidism have been described. The latter is less common, and until 1965 only 5 cases had been reported (Hayles, Hinrichs, and Tauxe, 1965).

The triple combination of hypothyroidism, Down's syndrome, and diabetes mellitus is even more rare and there are only two previously described cases (Daniels and Simon, 1968; Litman, 1968), who were aged 13 months and 17 years at the time the full picture became evident.

Investigations in this child have shown that she was hypothyroid rather than totally deficient in thyroid function. The presenting symptom of ascites is unusual in children, but fluid accumulation in the peritoneum and elsewhere is a well-recognized clinical feature of adult myxoedema. On admission the provisional diagnosis in this case was Mauriac syndrome (Guest, 1953), but it is doubtful if this condition of hepatomegaly, stunting of growth, and poorly controlled diabetes mellitus is an entity. Rather, this patient shows a rare cause of that unusual condition.

Summary

A child with Down's syndrome who subsequently developed diabetes mellitus and hypothyroidism before the age of 6 years is described. The principal presenting feature of her disturbed thyroid function was ascites.

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Cartilage hair hypoplasia

Cartilage hair hypoplasia was first described by McKusick in 1964. While studying the old Amish sect he noticed an association between short-limbed dwarfism and sparseness of hair, and later reported