

Correspondence

Congenital nephrogenic diabetes insipidus

Sir,

Schreiner and co-workers reported a baby girl with congenital nephrogenic diabetes insipidus (NDI) (*Archives*, 1978, 53, 906). I should like to contribute a case diagnosed as having partial NDI.

This boy had been transferred from a local hospital at age 9 months because of high fever with high serum Na and Cl concentrations, initially noticed in the first week of life. His mother showed polydipsia and polyuria: her urine osmolality after water deprivation for 12 hours was 270 mmol/kg. Investigation of the patient showed high serum osmolality (315 mmol/kg), low urine osmolality (100 mmol/kg), serum Na 172 mmol/l, and serum Cl 126 mmol/l.

Subsequently clearance studies were performed while the boy was receiving both a normal and a salt-restricted diet, after water deprivation, and after intravenous DDAVP (Aronson and Svenningsen, 1974). The main results were: (1) On water deprivation the patient was able to concentrate urine up to 600 mmol/kg simultaneously with reduction of urine volume; after DDAVP (0.4 ml IV) urine volume decreased further; osmolar clearance, which had increased during water deprivation, decreased after DDAVP. (2) On normal diet and on salt-restricted diet DDAVP resulted in increasing osmolar and free water clearance (and urine volume).

We made the following interpretations on the effect of DDAVP (a synthetic vasopressin analogue) on our patient's kidneys: renal resistance to DDAVP was incomplete; there was an adequate response after giving DDAVP during water deprivation, whereas a paradoxical effect on osmolar and free water clearance (Brodehl *et al.*, 1965) was apparent during the control periods.

Partial NDI (McConnell *et al.*, 1977) was diagnosed and treated by salt restriction and frusemide (15 mg/day). Follow-up 3 years later shows uncomplicated physical and psychological development of the patient with normal or almost normal values for serum Na, Cl, urea nitrogen, osmolality, and bicarbonate. Urine osmolality ranged from 200 to 265 mmol/kg.

I wonder if Schreiner and co-workers would like to comment on our patient, particularly with regard to their own therapeutic experience.

References

- Aronson, A. S., and Svenningsen, N. W. (1974). DDAVP test for estimation of renal concentrating capacity in infants and children. *Archives of Disease in Childhood*, 49, 654-659.
 Brodehl, J., Gellissen, K., and Hagge, W. (1965). Die Wirkung des Vasopressins beim Diabetes insipidus renalis. *Klinische Wochenschrift*, 43, 72-78.

McConnell, R. F., Jr, Lorentz, W. B., Jr, Berger, M., Smith, E. H., Carvajal, H. F., and Travis, L. B. (1977). The mechanism of urinary concentration in nephrogenic diabetes insipidus. *Pediatric Research*, 11, 33-36.

P. KOEPP
 Universitätskinderklinik,
 Martinistrasse 52,
 D-2000 Hamburg 20,
 W. Germany

Myotonic dystrophy and bonding failure

Sir,

In their report of 5 cases of the neonatal form of dystrophia myotonica (*Archives*, 1979, 54, 331), Pearse and Höweler emphasise the ethical problems facing the clinician once the diagnosis has been made, and they discuss the need for genetic counselling. We should like to add support to their concern for earlier diagnosis by drawing attention to another important and potentially lethal hazard run by babies with this condition—that is bonding failure. If they survive the neonatal period, these babies must always be considered to be at increased risk of rejection and neglect. Not only are they likely to have accumulated a number of the features commonly associated with child abuse—for example, neonatal separation, early ill health, maternal physical and emotional illness (Lynch, 1975; Lynch and Roberts, 1977)—but both mother and child are further handicapped by the inability to use facial expression effectively as a means of communication.

This was well illustrated by a family referred to the Park Hospital in Oxford. The parents were both very young and came from unhappy and divided homes, thus the potential for abuse was high. The mother's relationship with her own mother was hostile yet dependent, and the father had experienced a rigid authoritarian upbringing. When we met the mother she was just 20 and had had two caesarean sections within 13 months. Her second son had been very ill in the neonatal period, spending the first 7 weeks of his life in the special care baby unit. He had required assisted ventilation for some days. During his stay in the unit the diagnosis of myotonic dystrophy was made both in himself and his mother. A distaff family history of dystrophia myotonica was discovered, with the mother's own mother, the grandmother, and a great aunt suffering from cataracts and facial diplegia. It was also found that the elder brother had a milder form of the disease.

When finally discharged home, the baby had a floppy, expressionless face; he made none of the grimaces and little noises a normal baby makes. His abnormal cry was quiet and ineffective; his smile did not appear. Feeding was a nightmare. Much of his time he slept 'like a dead