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Fanconi's Syndrome with Renal Salt Wasting and Alkalosis

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Prof. J. A. Davis)

A 4-year-old boy with the non-cystinotic form of Fanconi's syndrome had all the usual clinical and biochemical changes but was unexpectedly *alkalotic*. A few other instances of this association have been reported, and the investigations were undertaken to explain this observation.

Addition of sodium chloride to the diet corrected the alkalosis, which recurred when the salt supplement was stopped. Further restriction of sodium intake enhanced the alkalosis, but urinary sodium wastage continued causing weight loss, depression of inulin and PAH clearances, and reduction of plasma volume. Aldosterone secretion rates were initially raised but fell after adequate sodium repletion of the patient.

When alkalosis was induced by dietary restriction of both sodium and potassium, sodium chloride replacement alone was only partially effective in correcting alkalosis: the addition of potassium salts was necessary for full correction. This produced a mild acidosis like that more commonly found in Fanconi's syndrome, and due to defective proximal tubular reabsorption of bicarbonate.

These observations throw light on the interrelation of alkalosis with the effects of contraction of the volume of extracellular fluid, chloride deficiency, and potassium depletion.

In the present case the tendency to bicarbonate loss was overcorrected by the kidney in its effort to conserve sodium; it differs from the usual case only in degree, by having large losses of sodium chloride rather than sodium bicarbonate.

Hydroxylysineuria

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Of 4 severely retarded sibs, 2, a boy and a girl, were found to excrete excessive urinary hydroxylysine. The

parents, born in 1929, are Anglo-Irish, unrelated, healthy, and of average intelligence. A younger sister of the propositi, born in 1962, is healthy and of average intelligence.

Case 1 (M.W., male, born in 1950). After normal pregnancy and delivery at 39 weeks weighing 3.2 kg. he developed neonatal jaundice of uncertain severity. Early progress was normal. He sat at 6 months and stood at 16 months. Shortly after this the first symptoms of trembling, hyperkinesis, myoclonic spasms, and major seizures were noted after an attack of measles. Development was arrested from this time. Epileptic fits ceased in early childhood. He has an IQ of 40, but no other clinical abnormality.

Case 2 (L.W., female, born in 1953). The pregnancy and delivery at full term were normal. Progress was normal until 7 months when she was noticed not to be using her left leg. At 19 months she developed trembling, myoclonus, hyperkinesis, and major seizures. Further progress ceased and speech was lost. At the present time she still has rare major epileptic attacks. She is plump, without speech, has an IQ of 30, but no other abnormality.

Case 3 (R.W., male, born in 1954). Pregnancy and delivery at full term were normal, birthweight 3.4 kg. He sat at 10 months, crawled and said single words at 1 year, stood with support at 16 months, but made no progress thereafter. Soon after his first birthday he became excitable, with unaccountable laughing and screaming, and developed tremors of brief duration which caused him to fall. Now he has an IQ of 25 but no other abnormality.

Case 4 (A.W., male, born in 1956). Birthweight 2.2 kg. after normal pregnancy and premature delivery. Apart from a tendency to vomit in early infancy, progress was normal (sitting at 10 months and crawling at 11 months) until it became arrested at 12 months when he developed tremors and infrequent major epileptic seizures, which lasted until the age of 8. Now he has an IQ of 25, no speech, but no other abnormal findings.

The majority of urinary hydroxylysine is present in peptides probably derived from degraded collagen. Normal values of total urinary hydroxylysine (assayed after acid hydrolysis of peptides) vary with age (range