of routine after the onset of 'paraplegia' in this group of patients.

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References

Geschickter C F & Maseritz I H
(1939) *Journal of Bone and Joint Surgery* **21**, 314–322
Lucas G L
(1978) *Clinical Orthopaedics* **137**, 85–86
Nanda S & Mohanti R C
(1968) *Journal of the Indian Medical Association* **51**, 185–186
Roth V G
(1976) *Clinical Orthopaedics* **117**, 247–253
Wilson J N
(1976) Watson-Jones' Fractures and Joint Injuries. 5th edn. Churchill Livingstone, Edinburgh; vol 2, p 1254
Zingraff J, Drueke T, Roux J P, Rondon-Nucete M, Man N K & Jungers P
(1974) *Clinical Nephrology* **2**, 73–75

Arginosuccinic aciduria with pili torti[1]

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Metabolic disorders associated with abnormalities of the hair are uncommon. We report a patient who had arginosuccinic aciduria and pili torti, a condition in which affected hairs are flattened and at three or four places along their shafts are twisted through 180°.

Case report

A female infant, born of Sephardic Jewish parents who were first cousins, was noted at birth to have an abnormally dark and greasy skin over the scalp. Subsequently, growth of the scalp hair, eyebrows and eyelids has been poor.

On examination, at the age of six weeks, there was sparse, stubbly hair over the scalp, and a scanty growth of eyebrow hair and eyelashes. The scalp was hyperkeratotic and there were several pustules scattered over it. The nails were normal. At nine months the scalp hair had improved a little. Mental and physical development have been otherwise normal.

Microscopy of the hair revealed pili torti. A scalp biopsy showed perifollicular chronic inflammation and fibrosis. Many of the hair follicles were abnormal and contained keratin plugs and misshapen hair shafts.

Examination of the parents and sibling did not reveal any abnormality.

Urinalysis by thin layer chromatography for amino acids revealed an unidentifiable 'spot' running between phenylalanine and alanine. Urine and plasma were then analysed by ion-exchange chromatography. Samples were deproteinized and acidified simultaneously using sulphosalicylic acid. Using a specific programme to allow separation from the basic column of arginosuccinic acid (ASA), homocystine, tyrosine, and phenylalanine, the patient's urine was found to contain two peaks which corresponded to the cyclic compounds formed when ASA is acidified and/or boiled (the anhydrides B and C of Westall 1966). Quantitation of the peaks was possible using the ninhydrin colour values quoted by Westall. In our patient's urine the quantity of compound B was 89 μmol/l (81 mg/g creatinine), and of C was 315 μmol/l (286 mg/g creatinine). In the plasma two peaks were found – one corresponding to pure ASA (252 μmol/l) and the other to anhydride B (259 μmol/l); anhydride C was not detected. ASA has been reported to be present in the blood of patients at levels of 10–400 μmol/l, and higher levels (400–1870 μmol/l) may occur in affected neonates (Shih 1978). A repeat urine analysis again showed anhydrides B and C in similar amounts.

Plasma and urine samples from the parents and one sibling showed very small, unquantifiable peaks of ASA in all samples but no anhydrides were detected.

Discussion

Arginosuccinic aciduria is a rare inborn error of metabolism. It is the result of a deficiency of an essential enzyme in the urea cycle – arginosuccinatelyase – and may lead to an increase in blood ammonia and citrullinaemia. Three clinical types have been described: (1) neonatal, with rapid deterioration and death in the first 10 days; (2) subacute, when feeding difficulties, failure to thrive and fits develop slowly in infancy; and (3) late-onset, in which neurological abnormalities and mental retardation become apparent in the second year. Whilst mental retardation is the rule, severe biochemical disturbance may occur in the absence of retardation, and asymptomatic siblings with arginosuccinic aciduria have also been reported (Applegarth *et al.* 1975, Shih 1978). The three clinical types of arginosuccinic aciduria are probably genotypically specific since the same type has recurred in each family. The available information suggests that the disorder is inherited as an autosomal recessive.

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About half of the severely affected patients with neurological disorders have abnormally friable hair, usually with trichorrhexis nodosa, and excrete several grams of ASA each day. However, minor hair abnormalities have been reported in otherwise normal patients who excrete traces of ASA: 10–20 mg/day (Winther & Bundgaard 1968, Shelley & Rawnsley 1966). At present, the hair abnormality cannot be correlated with the severity of the biochemical disorder and the basic biochemical defect in the hair remains unknown.

References

Applegarth D A, Davidson A G F, Perry T L, Poon S, Crichton J U & Hardwick D F
(1975) *Clinical Chemistry* **21**, 950–951
Shelley W B & Rawnsley H M
(1966) *Transactions of the Association of American Physicians* **47**, 146–156
Shih V E
(1978) In: Metabolic Basis of Inherited Disease. 4th edn. Ed. J B Stanbury *et al.* McGraw Hill, New York; pp 373–378
Westall R G
(1966) *Biochemical Journal* **77**, 135–144
Winther A & Bundgaard L
(1968) *Acta Dermato Venereologica* **48**, 567–570