Pediatr Nephrol (1996) 00: 000–000

Pediatric Nephrology

Brief report

Acrorenal syndrome associated with visual defect

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Received November 17, 1995; received in revised form and accepted June 11, 1996

Abstract. The clinical, biochemical, radiological, and histological data of a 5-year-old boy with severe limb deformities and renal failure due to oligomeganephronia and renal hypoplasia are reported. This patient represents another example of acrorenal syndrome. This boy has a severe visual defect due to pigmentory retinopathy, which has not been reported previously.

Key words: Acrorenal syndrome – Oligomeganephronia – Limb defects – Pigmentary retinopathy – Renal failure

Introduction

The association of limb deformities and renal malformations has been described in different reports under the term acrorenal syndrome [1–5, 7–9]. The renal anomalies vary from mild anatomical malformations to severe renal hypoplasia and renal failure [1]. The underlying renal pathology associated with renal failure has mainly been described as oligomeganephronia [4, 5, 7]. Presentation with end-stage renal failure is rarely reported [3–5]. We describe a male with end-stage renal failure, upper and lower distal limb deformities, and blindness due to pigmentary retinopathy; a finding which has not previously been described in this syndrome.

Case report

A 5-year-old boy with end-stage renal disease and congenital anomalies was referred to our unit from a regional hospital for further management. He was the product of a full-term uneventful pregnancy and delivery to a healthy 25-year-old mother and a healthy 55-year-old father with one normal sibling, a girl of 2 years of age. The parents were not consanguineous and there was no family history of skeletal or renal diseases.

Fig. 1. Photograph of the patient showing upper and lower limbs with severe deformities

Deformed hands and feet were noticed immediately after birth, but no major illnesses were reported in the first 3 years of life. Thereafter, he was noticed to be lethargic and pale. Investigations revealed renal failure and he was sent for further management.

On arrival, he was pale, his height and weight were below the 3rd percentile and his blood pressure was normal for age. He had bilateral jerky horizontal and vertical nystagmus with a severe defect in visual acuity and no coloboma; fundal examination by an ophthalmologist showed myelinated nerve fibres, pale discs, and darkly pigmented maculae. The left testicle undescended. Limb defects were obvious and are shown in Fig. 1. Intellectual development was appropriate for age. Laboratory investigations revealed: urea 40 mmol/l, creatinine 360 μmol/l, sodium 144 mmol/l, potassium 5.6 mmol/l, calcium 2.2 mmol/l, phosphorus 2.7 mmol/l, chloride 103 mmol/l, hemoglobin 72 g/l, and platelets 420 × 10⁹/l.

Abdominal ultrasonography showed a (hemoglobin) normal liver and spleen and small kidneys with high echogenicity. Bipolar measurement of the right kidney was 4.5 cm and left kidney 4.3 cm. Skeletal survey showed congenital reduction deformities of both feet with complete absence of tarsal bones except for talus and calcaneum. There was also complete absence of forefeet except for one rudimentary digit on the left side. Both hands showed congenital reduction deformities with only three metatarsals and one digit remaining on each side. Brain computed tomography showed minimal brain atrophy.

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Springer-Verlag, Heidelberg | PNE114
Mskr.-Seiten: 7 | Seiten: 2
Druckerei: Ernst Kieser GmbH, 86345 Neusäß | Versand:

Echocardiogram was normal. Auditory brain response and visual evoked potential were normal. Electroretinogram showed a normal response in both eyes, apart from diminished amplitude on the right side. Chromosomal analysis was normal (46, XY).

Left percutaneous renal biopsy showed a small number of glomeruli. The majority were sclerosed and hyalinized and the others were enlarged. The minimum glomerular diameter was 240 μm and there was proliferation of mesangial cells with mesangial matrix expansion. The interstitium showed focal hypercellularity and chronic inflammatory cell infiltrations, while the renal tubules showed focal atrophy with peritubular hyalinization. Many tubules also showed luminal dilation and irregular outline with degenerative changes.

Discussion

Acrorenal syndrome has been described by different observers since the detailed description of Curran and Curran [8], the severe limb deformities being common to all reported cases [1–4, 7–9]. The lower limbs were more severely affected than the upper limbs in the majority of cases described. Zaier et al. [5] reported an adult with acrorenal syndrome in whom the limb defects were more in the upper than the lower limbs. Renal involvement in their patient was not severe, as in other cases, and these authors postulated that severe lower limb defects may be associated with severe renal involvement. In our patient, the upper and lower limb defects were equally severe.

The reported renal involvement in acrorenal syndrome varies from mild hydronephrosis with duplex system to severe bilateral renal hypoplasia and renal failure. Renal pathology has not been frequently described, but the majority of patients who had renal biopsies had oligomeganephronia [4, 5, 7], as did our patient, and all developed renal failure. Oligomeganephronia is associated with extremely small kidneys [6]; the kidneys of our patient were also severely hypoplastic. Cryptorchidism and hypospadias are common associations with other renal anomalies and present in about 50% of males with acrorenal syndrome [5, 8]. Our patient had an undescended left testicle and required orchiopexy.

Visual impairment was obvious in our patient. Fundal examination revealed pigmentary retinopathy, a unique finding which, as far as we know, has not been reported before. Buttiens and Fryns [7] reported a 16-year-old boy with limb deficiency and proteinuria due to oligomeganephronia with severe myopia (–12 dipters), but did not describe fundal examinations.

The previous speculation of X-linked recessive inheritance based on the predominance of males is rendered less likely by the recent observation of the syndrome in girls [4, 7]. We agree with others [8, 9] that high paternal age, as in our patient, could be a risk factor; the mode of inheritance in this case fits more an autosomal dominant pattern or a germline mosaicism for a dominant gene.

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