
Communications

The Genogram as an Aid to Diagnosis of Distal Renal Tubular Acidosis

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The genogram, a graphic display of kinship through multiple generations, has been described by Guerin.¹ Antedated by similar concepts such as the pedigree chart,² the genogram was described in detail in a previous report by Jolly et al,³ who dealt with the mechanics and symbols used in its construction. The genogram concept has been taught and used for a number of years in the University of South Alabama Department of Family Practice. A genogram is routinely obtained on all patients and becomes a permanent document in their record.

Because of its graphic qualities and its ability to be obtained easily, the genogram helps to derive important clinical information. With a systematic approach, a complete medical, genetic, social, and psychological assessment can be made quickly and efficiently.

Renal tubular acidosis is a distinct clinical syndrome associated with growth failure, abnormal skeletal growth, and metabolic acidosis. Primary distal renal tubular acidosis generally becomes manifest during infancy or early childhood. It has been shown to have a familial pattern of inheritance, generally transmitted as an autozomal dominant trait with variable penetrance.^{4,5}

This case report illustrates the usefulness of the genogram in diagnosing renal tubular acidosis in an eight-year-old child.

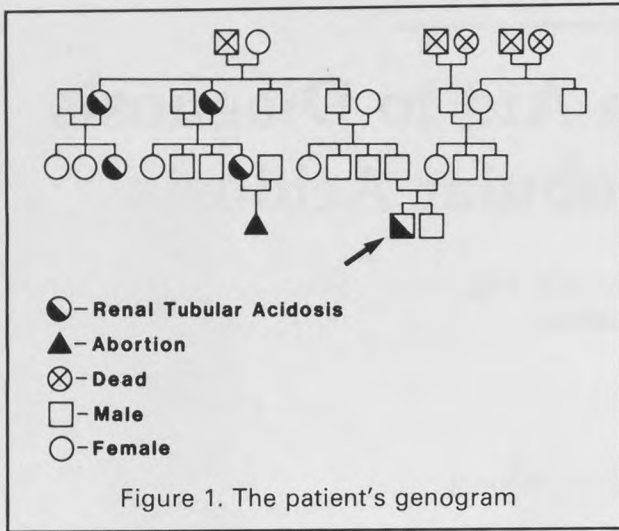
Case Report

The patient was an eight-year-old white boy who had a normal birth history and a normal birth weight and length. He was noted over several years, however, to be growing poorly despite adequate appetite and activity. The patient had a male sibling two years younger than he who had normal growth. When the patient was five years of age, his parents noted that he had decreased energy and had developed a nervous habit of rubbing his fist on his face and grinding his teeth. At six years of age he was hospitalized by another physician for growth failure and evaluated for hypopituitarism. The boy was found to have normal results on an arginine-insulin infusion test with normal serial growth hormone and normal thyroid function levels. Bone age films at that time revealed the patient's bone age to be three years, nine months. Skull roentgenograms were normal. A specific diagnosis was not made at that time.

The patient subsequently began to manifest genu valgum and was given corrective shoes. He did not enjoy playing with friends because he could not run well. He also developed polydipsia and nocturia.

On his second admission at eight years of age, a genogram was obtained (Figure 1) significant in that it revealed four relatives, two paternal great aunts and one child of each, who were affected with renal tubular acidosis. Given this distant family history of renal tubular acidosis, the same diagnosis was entertained in this patient. Arterial blood gases were obtained revealing a pH of 7.29, PaO₂ of 111 mmHg, PaCO₂ of 26 mmHg and a bicarbonate of 12 mEq/L. The urine pH under oil was 6.5. Electrolytes revealed sodium 135 mEq/L,

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potassium 3.1 mEq/L, chloride 113 mEq/L, and bicarbonate 12 mEq/L. These data are consistent with the diagnosis of renal tubular acidosis. The child was begun on modified Shohl's solution. Since institution of the appropriate therapy, the child has made dramatic growth improvement and presently is above the 10th percentile for his age. On 12.5 mL of Shohl's solution four times daily his serum bicarbonate is normal.

Comment

This case illustrates the usefulness of obtaining a genogram as a routine part of the patient's history. In this particular patient a diagnosis had been delayed because it was not known that the child had distant relatives with renal tubular acidosis, and the diagnosis was not suspected. In this and in a number of other cases seen in which a diagnosis either was not suspected or was suspected, but difficult to confirm, the genogram has been invaluable. Consequently, it is advocated that a genogram be obtained for every patient, especially for those patients who pose difficult diagnostic problems.

References

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Teaching Family Practice Residents to Identify and Treat Battered Women

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In recent years the medical and psychological problems of battered women have come to the attention of the medical community. Current research places the degree of the abuse as occurring

in one out of six couples.¹ The startling numbers of physically abusive relationships have triggered a response in the medical community. Protocols for emergency room treatment of abuse have been developed at hospitals and medical schools, often with the cooperation of community agencies who help battered women.²⁻⁵ However, the recognition of abuse in a non-emergency-room setting and the implementation of appropriate response in the family practice setting have not been addressed.

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