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Male pseudohermaphroditism with 17 alpha-hydroxylase deficiency. A case report.

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Abstract

A case of male pseudohermaphroditism with 17 alpha-hydroxylase deficiency is reported in a 23-year-old woman presenting with primary amenorrhoea and a history of bilateral inguinal hernia repair. She was tall, had hypoplastic external genitalia with a blood pressure of 220/140 mm Hg. Her karyotype was XY. Acute adrenal failure occurred following exploratory laparotomy. After treatment with glucocorticoids and oestrogens, the hormone and electrolyte profiles returned to normal and the blood pressure fell. The biochemical implications of this enzyme deficiency are discussed.

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