Pituitary Function in a Patient with Septo-Optic Dysplasia and Pituitary Dwarfism (Kaplan-Grumbach-Hoyt Syndrome)

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Abstract. The case of a seventeen-year-old female patient with septo-optic dysplasia and pituitary dwarfism is presented. Mental retardation and epilepsy, in addition to absence of the septum pellucidum, point to a widespread lesion of the central nervous system. There is unilateral hypoplasia of the optic nerve. She is of small stature. The dynamic pituitary tests point to deficiency of GH, TSH and ACTH, and an adequate reserve of prolactin, gonadotrophins and vasopressin. TSH insufficiency is probably of primary pituitary origin.

Key words: Multiple congenital anomaly (MCA) syndrome – Dwarfism – Hypopituitarism – Secondary hypothyroidism – Releasing hormones.

Introduction

The pathologist DeMorsier (1956) described septo-optic dysplasia as a clinical entity which includes: (1) lack of the septum pellucidum; (2) a primitive optic ventricle; (3) hypoplasia of the optic nerve, chiasma and the optic tract. Hoyt et al. (1970) and Kaplan et al. (1970) have described six patients with sporadic hypopituitary dwarfism and optic nerve anomalies. In four of these children, pneumoencephalograms were taken and in three of them the absence of the septum pellucidum was established. Since the publication of this description septo-optic dysplasia and pituitary dwarfism is considered a syndrome including pituitary dwarfism, hypoplasia of the optic nerve and malformation of the prosencephalon. Further cases were described by Ellenberg and Runyan (1970), Brook et al. (1972), Harris and Haas (1972), Billson and Hopkins (1972), Patel et al. (1975) and Toublanc et al. (1976).

We have studied a girl in whom all the features of the syndrome of septo-optic dysplasia and pituitary dwarfism were present. Our observations are presented as...
a contribution to the elucidation of the pathogenesis of hypopituitarism in this syndrome.

**Case Report**

The girl was born in 1958 and at the time of examinations she was 17 years of age. She was the first child of healthy parents and there is no consanguinity in the family. No other members of the family have abnormally short stature or suffer from an endocrine disorder. In the third month of the pregnancy, the mother had a mild viral infection. The girl was born at term and the delivery was normal. The infant weighed 4300 g and was 52 cm long. Soon after birth, convergent strabismus and hemiparesis of the left arm and leg were noted. At the age of 4 years the girl developed generalized convulsions. At that time it was also established that she could not see with her left eye. The convulsions could be kept under control with Hydanphen. As a young child she was smaller than girls of her own age and her abnormal short size became especially conspicuous after the age of seven years.

The girl is abnormally short with a relatively normal body weight. She is very placid and slow, and also speaks very slowly. Height 143 cm (-3.5 SD), span 145 cm, and upper lower segment ratio 0.82 (normal 1.03). The hands and feet are small and the head circumference is 50 cm. Weight 36.5 kg (-3.5 SD). The tone and strength of the muscles of the left arm and left leg are reduced. The temperature of the skin of the left extremities is 1°C lower than the skin on

![Fig. 1. Lumbar air encephalogram: AP view of the skull showing air in a single midline anterior cerebral ventricle, and absence of the septum pellucidum](image)