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Septo-optic dysplasia and congenital panhypopituitarism associated with complicated gastroschisis

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ABSTRACT

Gastroschisis is a rare defect of the abdominal wall, rarely associated with other malformations. Septo-optic dysplasia (SOD) is a midline abnormality characterized by hypothalamic-pituitary dysfunction and cerebral parenchymal abnormalities, both of unknown causes.

I present the case of a preterm newborn with a weight appropriate for her gestational age and a prenatal diagnosis of gastroschisis. Surgical repair was performed 8 hours after birth. Subsequently, she developed fluid and electrolyte imbalances and polyuria; central diabetes insipidus was diagnosed based on a positive desmopressin test. A brain MRI was performed, showing findings consistent with SOD, and hormonal studies confirmed congenital panhypopituitarism. Hormone replacement therapy was initiated, and the patient's clinical course has been favorable.

SOD and congenital panhypopituitarism are difficult to diagnose due to their variable and nonspecific presentation. Hypoglycemia is the predominant finding in newborns. Early detection allows for treatment and prevents irreversible complications.

Keywords: *septo-optic dysplasia; congenital hypopituitarism; gastroschisis; neurogenic diabetes insipidus.*

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INTRODUCTION

Gastroschisis is one of the most common congenital defects of the abdominal wall[1] and is rarely associated with other malformations. Ultrasound usually diagnoses it prenatally between the 12th and 16th weeks of gestation.

Septo-optic dysplasia (SOD) is a heterogeneous anatomoclinical midline malformation with an unknown incidence; it is estimated to affect about 10 out of every 100 000 live births.[2] It is characterized by hypoplasia of the optic nerves and the corpus callosum, total or partial absence of the septum pellucidum, variable dysfunction of the hypothalamus and pituitary gland, and abnormalities of the cerebral parenchyma. Clinicians typically establish the diagnosis by 12 months of age, based on growth failure secondary to growth hormone deficiency and visual impairments associated with developmental delay.[2,3]

Early diagnosis of SOD allows for timely initiation of hormone replacement therapy, prevents growth and neurodevelopmental abnormalities, and enables early initiation of stimulation therapies. The objective of this study is to describe the clinical case of a patient with a prenatal diagnosis of gastroschisis who had a difficult postoperative course due to panhypopituitarism associated with SOD. This study describes the disease's main characteristics and the importance of early diagnosis.

CLINICAL CASE

A preterm infant at 36 weeks' gestation with a birth weight of 2610 g, length of 45 cm, and head circumference of 32 cm. The infant was delivered by cesarean section due to premature rupture of membranes. The pregnancy was diagnosed at 16 weeks' gestation, with five subsequent follow-up visits confirming the presence of gastroschisis via ultrasound and magnetic resonance imaging. The mother is 20 years old, and the father is 19 years old; both are healthy, with no consanguinity, and this is their first child. Family history: maternal and paternal grandmothers were healthy; the paternal grandfather had hypertension; and the maternal grandfather was healthy. During the first trimester of pregnancy, the mother reported daily consumption of alcohol, tobacco, and marijuana; serology tests were negative.

At birth, the newborn showed no dysmorphic features. Gastroschisis complicated by a restrictive parietal defect was diagnosed, and the infant's anthropometric measurements were appropriate for the gestational age.

At 8 hours of life, she was referred to the Hospital de Niños Ricardo Gutiérrez, where the gastroschisis was surgically closed without complications. Twenty-four hours later, she developed polyuria and hypernatremia that were difficult to manage, with sodium levels ranging from 127 to 168 mEq/L (*Table 1*). On physical examination, she was eucardiac, eupneic, and hemodynamically stable, with generalized hypotonia, poor responsiveness to her surroundings, and weak crying and sucking reflexes; no other relevant findings were noted.

During correction of hypernatremia with free water, the patient experienced seizures, which were interpreted as symptomatic hyponatremia secondary to a sudden drop in serum sodium (29 mEq/L in less than 48 hours). Renal function was normal, and the absence of proteinuria in the 24-hour urine collection ruled out a renal cause for the fluid and electrolyte disturbance.

At 9 days of age, polyuria and hypernatremia persisted. The desmopressin test showed a good response. Although serum sodium stabilized and

Table 1. Serum sodium levels

Age	Sodium level (mEq/L)
8 hours of life	128
3 days of life	163
5 days	127
6 days	146
9 days	161
11 days	139
12 days	158
13 days	142
15 days	169
19 days	154

urine output improved, the patient developed lethargy and fluctuating consciousness and required high glucose in parenteral nutrition to maintain normoglycemia (*Table 2*). Given the clinical diagnosis of central diabetes insipidus, supported by elevated plasma osmolarity (332 mOsm/kg; normal range: 280-295 mOsm/kg) and a favorable response to desmopressin, a central endocrine disorder was suspected.

Hormone testing revealed multiple hypopituitarism with hypothyroidism (elevated thyroid-stimulating hormone [TSH] and decreased free thyroxine [FT4]), decreased adrenocorticotropic hormone (ACTH) with low cortisol, and deficiencies in growth hormone and sex hormones (*Table 3*). Levothyroxine, hydrocortisone, and somatotropin improved sensory function and achieved adequate glycemic control.

At 21 days of life, we performed a brain MRI. Findings included reduced-diameter optic nerves, an absent pituitary stalk, an absent neurohypophysis in its usual location, hypothalamic dysgenesis, a hypoplastic optic chiasm, and an absent septum pellucidum (*Figure 1*). These findings, together with the endocrinological profile and clinical presentation, led to a diagnosis of SOD with congenital panhypopituitarism and central diabetes insipidus in a patient with complicated and corrected gastroschisis.

During the hospital stay, we performed additional tests to rule out other malformations. Fundus examination revealed pigmented margins of both optic discs, left optic disc hypoplasia, and a deep temporal hemorrhage in the right eye; visual acuity was limited to hand motion. The echocardiogram revealed a patent ductus arteriosus and a patent foramen ovale without hemodynamic impact, with preserved ventricular function, consistent with a normal finding for the gestational age. Abdominal and renal Doppler ultrasound showed no abnormalities.

The patient's condition improved following the initiation of hormone therapy, with adequate weight gain, normalization of serum sodium levels and diuresis, and resolution of neurological symptoms.

She is currently undergoing interdisciplinary follow-up at our center with the High-Risk Pediatrics, Ophthalmology, Neurology, and Endocrinology departments, and her growth and neurodevelopment are consistent with her corrected age. Because she has two congenital malformations, the Genetics Service evaluated her and began monitoring her case. During her hospitalization, no karyotyping or genomic testing was performed; these were subsequently ordered on an outpatient basis, which limits our ability to establish the etiology of SOD in this patient.

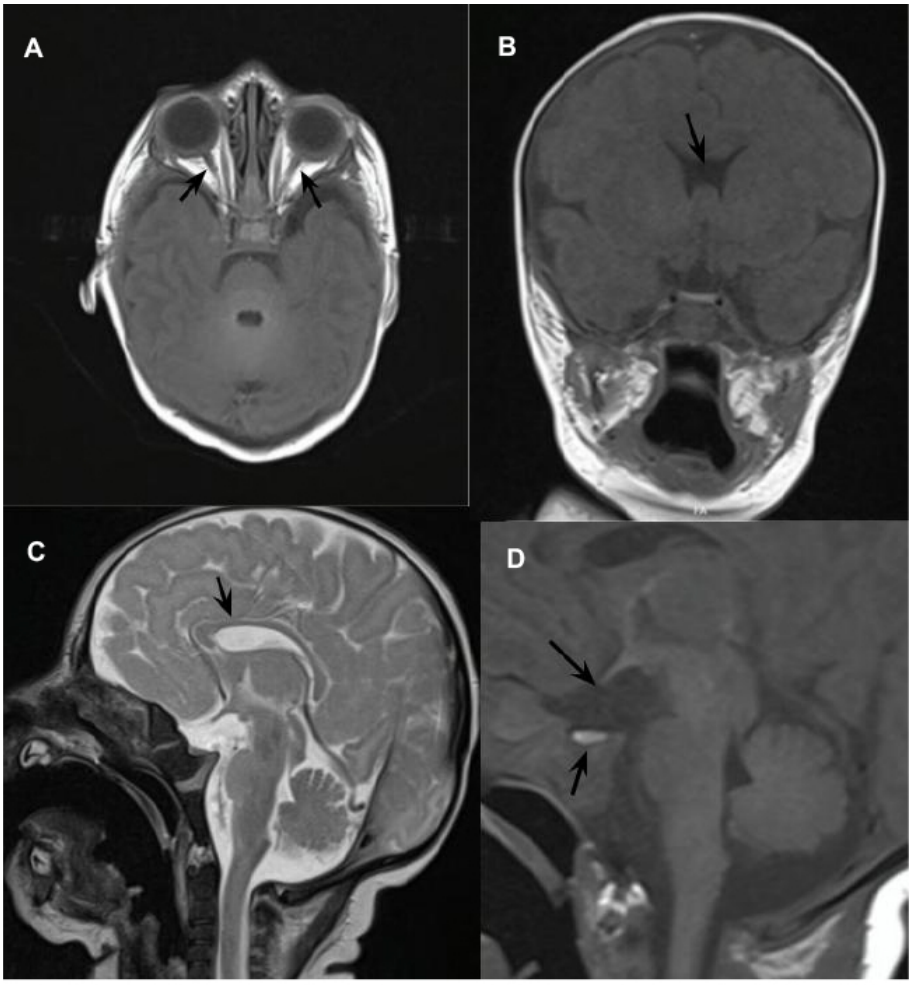
Table 2. Blood glucose levels

Age (days)	Blood glucose (mg/dl)
5	77
6	31
8	60
14	43
18	59
21	52

Table 3. Endocrinology test laboratory values (14 days of life)

Test	Patient's value	Normal range for age
Thyroid-stimulating hormone	5.27 µU/ml	1-2.6 µU/ml
Free thyroxine	0.67 ng/dl	1-2.6 ng/dl
Adrenocorticotropic hormone	5 pg/ml	10-46 pg/ml
Cortisol	0.3 µg/dl	1.2-15.1 µg/dl
Growth hormone	3.72 ng/ml	>6.5 ng/ml
Prolactin	196.1 ng/ml	43-331 ng/ml
Luteinizing hormone	0.1 mUI/ml	0.10-1 mUI/ml
Follicle-stimulating hormone	0.3 mUI/ml	0.3-8.7 mUI/ml

Figure 1. Magnetic resonance imaging of the central nervous system



A: small-diameter optic nerves and hypoplastic optic chiasm. B: absence of the septum pellucidum. C: thinning of the corpus callosum. D: absence of the pituitary stalk, absence of the neurohypophysis in its normal location, and hypothalamic dysgenesis.

DISCUSSION

SOD associated with congenital panhypopituitarism poses a diagnostic challenge during the neonatal period due to the variability and nonspecific nature of its clinical manifestations. The most common presentation is treatment-resistant hypoglycemia secondary to hypopituitarism.[4] Cases have been reported of infants who presented during the first three months of life with hypoglycemia and hypocortisolism, progressed to severe decompensation, and were subsequently diagnosed with SOD.[5-7] The remaining hormonal deficiencies usually manifest later; short stature is the primary reason for medical consultation.[2,3]

The most common manifestations include ocular abnormalities such as nystagmus and decreased visual acuity, followed by neurological involvement, primarily seizures, and hormonal deficiencies secondary to hypopituitarism.[3,4] Central diabetes insipidus is rare in the neonatal period; however, it has been reported in up to 25% of patients with SOD. Growth retardation and, occasionally, hypoglycemia occur in approximately 70% of cases, and the average age at presentation is 5 years.[2,3] Neurological manifestations are variable and may be related to associated cortical abnormalities, with seizures, focal deficits, and severe developmental delay observed.[3] In the absence of cortical abnormalities, patients may be asymptomatic or exhibit cognitive deficits secondary to thyroid

hormone deficiency. In our patient, suspicion of central diabetes insipidus—given a difficult postoperative course with difficult-to-manage hypoglycemia and neurological involvement—led to a diagnosis of SOD and panhypopituitarism before she was one month old. Early initiation of hormone replacement therapy was associated with growth and neurodevelopment consistent with corrected age. Although gastroschisis is the most common congenital abdominal wall defect, it rarely coexists with other malformations.[1,8] The coexistence of gastroschisis and SOD has only been reported in isolated cases; the available evidence does not allow for establishing a causal association between the two conditions. Both gastroschisis and SOD have an etiology that remains unclear and have been linked to exposure to toxins during the first trimester of pregnancy and to vascular anomalies.[1,3]

Two etiological hypotheses have been proposed for SOD.[9] The vascular hypothesis posits an abnormality in the development of the anterior cerebral artery, which would cause hypoplasia of the optic nerve and/or the septum pellucidum, with subsequent extension to the pituitary gland and the cerebral cortex.[9] The genetic hypothesis describes mutations in *HESX1*, located on the short arm of chromosome 3 and responsible for prosencephalization during embryonic development.[3,9] In addition, mutations in *OTX2*, *SOX2*, and *SOX3*—genes involved in the embryonic development of the optic nerve and the adenohypophysis—have been implicated.[10] However, variants in *HESX1* are identified in only about 10% of cases, suggesting the involvement of other genes that have not yet been fully characterized.[3,10] Most cases of SOD are attributed to de novo mutations, although hereditary forms have also been described.[11]

Early diagnosis of SOD is essential because of its frequent association with multiple hypopituitarism and the need for early initiation of hormone replacement therapy, particularly with thyroid hormones to prevent neurodevelopmental abnormalities and with growth hormone to prevent growth failure. Because this rare condition is difficult to recognize in the neonatal period, consider it when symptoms suggest pituitary hormone deficiency to establish a timely diagnosis and initiate early treatment. ■

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