A neonate with gastroschisis and hydrocephalus complicated by central diabetes insipidus

Timothy Dribin, Ryan M. McAdams

Department of Pediatrics, University of Washington and Seattle Children’s Hospital, Seattle, WA, USA

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Abstract

Gastroschisis is typically considered an isolated anterior abdominal wall defect with a low rate of secondary gastrointestinal anomalies. However, other congenital anomalies associated with gastroschisis may be more common than is generally appreciated. We report a case of term neonate with gastroschisis and congenital hydrocephalus who developed central diabetes insipidus. This case highlights the complexity of managing neonates with central diabetes, which often requires multiple pharmacologic agents such as vasopressin and thiazide diuretics. Although rare, assessing patients with gastroschisis for other anomalies is important since conditions like hydrocephalus may complicate medical management.

1. Case

A 3400 g male with prenatal ultrasound diagnosis of gastroschisis and hydrocephalus was delivered without complications by planned cesarean section at 38 weeks’ gestation because of macrorhaly. An initial prenatal ultrasound at 27 weeks’ gestation identified mild ventriculomegaly and an abdominal wall defect to the right of the umbilical cord insertion site consistent with gastroschisis. A 34-weeks’ gestation ultrasound demonstrated that the abdominal wall defect was unchanged with normal bowel caliber and no evidence of obstruction; however, there was severe ventriculomegaly with new dilation of the third ventricle consistent with, but not specific for aqueductal stenosis. The patient’s mother had genetic analysis with cell free DNA, which was normal. She declined amniocentesis for cytogenetics or microarray because it would not change her management of the pregnancy. After birth the patient was transferred to our institution for surgical repair of his gastroschisis. His exam was significant for an anterior abdominal wall defect consistent with gastroschisis, and an enlarged occipital-frontal circumference (40 cm, 100th percentile), without any facial dysmorphisms suggestive of an underlying syndrome. He was found to have distal ileal atresia and subsequently underwent uncomplicated closure of the gastroschisis with end ileostomy and ileal mucous fistula.

A cranial ultrasound on day of age one demonstrated moderate ventriculomegaly (3 mm) with mild dilatation of the lateral and third ventricles. A sequential ultrasound showed increasing midline shift bilaterally consistent with hydrocephalus. A subsequent MRI showed severe hydrocephalus of the lateral and third ventricles with cerebral aqueductal obstruction as well as partial lissencephaly and small bilateral optic pathways without evidence of septo-optic dysplasia or an intracranial tumor (Fig. 1). A screening renal ultrasound and echocardiogram were normal. The infant had no apneic, bradycardic, or cyanotic events and did not require any respiratory support.

On day of age 2, the infant developed increasing urine output of 4.5 ml/kg/hr with a total fluid requirement of 175 ml/kg/day. He had
not received any diuretic therapy. Laboratory analysis supported the diagnosis of diabetes insipidus based on an elevated sodium level of 162 mEq/L, serum osmolality of 303 mOsm/kg, and urine osmolality of 269 mOsm/kg. The endocrinology team was consulted and the infant was started on volume replacement to compensate for insensible losses due to polyuria (1:1 replacement with 1/2 normal saline to match urine output) together with his total parenteral nutrition maintenance fluids, which were held constant to ensure the infant received adequate nutritional intake despite fluctuating fluid requirements from his CDI. Additionally, he was also started on a vasopressin drip to maintain stable urine output and sodium levels. On day of age 3, the vasopressin drip was increased because his urine output was elevated to 16 ml/kg/hr requiring total fluids of 520 ml/kg/day.

The CDI was presumed secondary to hydrocephalus. The infant underwent endoscopic third ventriculostomy with bilateral choroid plexus cauterization and placement of a ventricular reservoir. Following the procedure his urine output and sodium levels normalized presumed secondary to increasing endogenous antidiuretic hormone secretion. His vasopressin drip was weaned off and he was transitioned to intravenous chlorothiazide per endocrinology recommendations. The chlorothiazide was titrated to a dose of 5 mg/kg twice daily to achieve normal urine output and sodium levels. The following week he was weaned off the chlorothiazide.

Biweekly cranial ultrasounds demonstrated stable ventricular size indicating effective cerebrospinal fluid drainage. However, after one month the infant developed cerebrospinal fluid leakage from the surgical site with increasing ventriculomegaly on cranial ultrasound indicating failure of the endoscopic third ventriculostomy and choroid plexus cauterization. He subsequently underwent uncomplicated removal of the ventricular reservoir with placement of a ventriculoperitoneal shunt since he was not a candidate for a ventriculoperitoneal (VP) shunt given his recent history of abdominal surgery. Around the time of his shunt placement, he again developed symptoms consistent with CDI based on high serum osmolality and sodium levels with low urine osmolality and sodium levels. He was started on daily enteral free water replacement and his laboratory values normalized.

Ophthalmological evaluation demonstrated bilateral optic nerve hypoplasia with concern for severe vision loss. A genetics consult did not lead to a unifying cause of his anomalies and a single nucleotide polymorphism array study was normal. DNA was banked for potential future analysis. The infant was slowly advanced from nasogastric tube gavage feeds to expressed breast milk nipple feeds of 170 ml/kg/day plus 20 ml/kg/day of free water and was discharged home at 44 weeks’ corrected gestation age.

2. Discussion

Non-intestinal anomalies are common in infants with omphaloceles, but are rarely reported in infants with gastroschisis [4]. Recent literature suggests that associated anomalies should be considered in gastroschisis cases. A retrospective analysis of 3,806,299 Texas births from 1999 to 2008 comparing the prevalence of associated anomalies among patients with gastroschisis and omphaloceles, found that 32% of patients with gastroschisis had associated anomalies versus 80% with omphaloceles [2]. The most common anomalies associated with gastroschisis included urogenital (24%), musculoskeletal (21%), and cardiac (15%) defects. Only 7.2% of infants with gastroschisis had anomalies that involved the central nervous system of which 1.2% had hydrocephalus [2]. We report a rare case of gastroschisis with hydrocephalus further complicated by CDI without evidence of an underlying genetic abnormality based on single nucleotide polymorphism array. In this case, we speculate that resolution of the CDI was likely secondary to treatment of the hydrocephalus with third ventriculostomy, as evidenced by stable cranial ultrasounds with diminishing need for vasopressin and chlorothiazide following surgical intervention. The mechanism by which hydrocephalus causes CDI remains unknown, but may be related to elevated intracranial pressure.

Literature describing an association between hydrocephalus and CDI in neonates is sparse. One case report documented a preterm neonate who developed intraventricular hemorrhage and post-hemorrhagic hydrocephalus who subsequently developed CDI requiring intranasal desmopressin [5]. Management of CDI in infants can be challenging and early consultation with pediatric endocrinology is recommended with close monitoring of urine output and sodium levels. Infants with CDI excrete excessive quantities of very dilute, but otherwise normal urine, resulting from impaired synthesis and secretion of antidiuretic hormone by the hypothalamus and posterior pituitary. Vasopressin is an appropriate initial therapy for central diabetes insipidus with transition
to chlorothiazide once the urine output and sodium levels have stabilized [6]. Although the exact anti-diuretic mechanisms in the setting of CDI are not known, chlorothiazide, a thiazide diuretic, inhibits the sodium-chloride co-transporter in the renal distal convoluted tubule and is thought to have a paradoxical antidiuretic action related to increased renal sodium excretion [7]. Increased sodium loss produces extracellular volume contraction leading to decreased glomerular filtration rate and increased proximal tubular sodium and water reabsorption.

Along with medical management directed at maintaining normal urine output and sodium levels, CDI in patients with hydrocephalus may be treated with neurosurgical intervention with a shunt or ventriculostomy. Although VP shunt placement is a common treatment for hydrocephalus, literature is lacking on the utility of VP shunts in patients with gastroschisis. Following gastroschisis repair, a number of factors could limit the effectiveness of a VP shunt including the technical challenge of placing a VP shunt into an underdeveloped abdominal wall cavity filled with swollen bowel, a risk for leakage through the gastroschisis repair site, and an increased risk for fluid pockets and pseudocyst formation due to abdominal cavity inflammation.

Thus, for patients with gastroschisis and hydrocephalus who fail endoscopic third ventriculostomy and choroid plexus cauterization, ventricular atrial shunts are likely the most appropriate treatment.

A high index of suspicion may be required to identify other anomalies in newborns with gastroschisis, but detection may be augmented by a screening ultrasound of the brain, heart, and kidneys. Infants with gastroschisis complicated by hydrocephalus should be closely monitored for the development of CDI and may require neurosurgical intervention along with medical management to correct fluid and electrolyte imbalances.

Conflict of interest
The authors have no conflict-of-interest, financial support, or other potential conflicts of interest to declare.

References