The Unusual Story of an Infant with Congenital Adrenal Hyperplasia

A one-week-old baby was brought to our outpatient department by her father and grandmother, with the complaint of ambiguous genitalia. She was the first offspring of a non-consanguineous couple from low socioeconomic strata. The baby was lethargic, hyperpigmented, and had clitoromegaly and partial labial fusion. Baby was hospitalized with provisional diagnosis of congenital adrenal hyperplasia (CAH). Blood test revealed acidosis, hyponatremia and hyperkalemia. Further investigations confirmed the diagnosis: ultrasonography showed ovaries and mullerian structures, serum 17-hydroxy progesterone (17OHP) was >200 ng/mL, and karyotype was 46,XX. Both parents were educated till middle school level. They were explained that the baby was a girl with normal internal reproductive organs; genitoplasty would be required to correct the ambiguity; and regular medication and follow-up were required. The baby was discharged on oral hydrocortisone, fludrocortisone and salt supplement.

After 10 days of discharge, the baby was again brought to our emergency services, with complaints of vomiting, lethargy, refusal to feed and respiratory distress. She was in hypovolemic shock. This time the baby was brought as a new case with a new name by a *kinnar* (transgender), who identified herself as the legal guardian of the baby. She carried an affidavit signed by both parents that the baby had been given to her willingly. The *kinnar* told us that she and her group were taking very good care of the baby. However, they had not been told about the need for regular medicines by the parents, and therefore the baby had not received the steroid supplementation for the last week. We called up the father (taking his telephone number from admission records), and asked him about the child. On his replying that the baby was well, we confronted him with the facts and called both parents to the hospital. The mother stayed with the baby and did seem to have an emotional bonding with the baby, but both parents were very young, and completely dependent on the paternal grandparents. Three senior consultants from Pediatrics and two from Pediatric Surgery were involved in the counseling. We spoke to the family multiple times, to the mother, father, and both sets of grandparents, singly as well as together, in tones ranging from coaxing to authoritative, and explaining to them scientifically as well as socially, invoking their conscience, giving a very positive outlook for the child and telling them about how well the girls with CAH in our follow-up were doing. Considering that ambiguity was the main hurdle to the parents accepting the baby, our pediatric surgeons agreed to carry out an early genitoplasty for the baby prior to discharge from the hospital. The parents and grandparents, however, kept changing their stand, agreeing to keep the baby on one day, and declining on the next.

After hospitlization for about three weeks, we were convinced that the family would not take care of the baby and in all probability, will either give away the baby to the *kinnars* or do something even worse. In consultation with our medical social service officer, the parents were asked to legally relinquish the baby to child welfare unit. The baby was then placed in a government-recognized orphanage. She was brought to OPD regularly by the caregivers and it was heartening to observe her steady growth and development over the next 9 months. Finally, the child has been legally adopted by an American couple, who according to their written submission, had already initiated enquiries for CAH support groups and Pediatric endocrinology and surgery facilities in their city, and were looking forward to welcoming the baby as their child.

For a patient with CAH, the outlook for successful sexual and reproductive life in adulthood depends upon the endocrine control and the cosmetic and functional results of genitoplasty [1]. Our case serves to bring to the fore the plight faced by numerous infants with ambiguous genitalia. Previously too, we have encountered families who never returned for follow-up after the initial diagnosis. Many of our CAH patients' parents have informed us about the social stigma faced by them and the child, from within their extended families and from peers [2]. Few have even had to separate from their joint families to be able to take care of their children. Many of them have had to rebuff attempts made by *kinnar* groups to force them to give away their children [2].

There is an urgent need to have patient support groups for newly diagnosed children with CAH. Additionally, a registry of children with CAH should be maintained at

national or state level, preferably along with implementation of universal newborn screening for CAH, so that affected individuals can be tracked and not allowed to slip between fault lines. Meanwhile, pediatricians who see these patients for the first time should also record their phone numbers and addresses, and ensure that the patients remain in follow-up. For families that are completely unwilling to come to terms with the diagnosis, the option of legal relinquishment and placement of baby in a recognized adoption agency may be exercised as a last resort.

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***VANDANA JAIN AND MADHULIKA KABRA**
Department of Pediatrics, All India Institute of Medical Sciences, New Delhi, India
**drvandanajain@gmail.com*

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Evisceration of Gut Through Post-surgical Drain Site in a Neonate

Intra-abdominal drainage following routine abdominal surgery continues to be a controversial subject [1]. The evisceration of various organs through drain site has been commonly reported in adults but is rarely seen in neonates. We report a case of small bowel loop evisceration out of the drain site in a 9-day-old neonate who was operated for duodenal atresia.

A newborn boy was admitted to the neonatal intensive care unit (NICU) of this hospital because of antenatal detected double bubble appearance in antenatal ultrasound of mother. At birth, the child weighed 2275 g and appeared vigorous, with normal spontaneous respiration. *X*-ray abdomen showed double bubble appearance in upper abdomen. Gastrograffin dye study confirmed the diagnosis of duodenal atresia. On day-2 of life, laparotomy was performed and duodeno-duodenostomy was done. An abdominal glove drain was brought out through a right iliac fossa stab incision approximately 8 mm. The intraoperative course was uneventful and child was shifted back to Neonatal medical unit. The patient responded well in the postoperative period. Feeding was started gradually from 5th post operative day. The drain was removed on the sixth postoperative day. On the 7th post operative day, a loop of small bowel prolapsed through the drain site while he was crying (***Fig.*1**). The child was shifted to operation theater. The prolapsed gut through the drainage site was healthy, slightly congested. It was reduced after enlarging the wound, and the drain site was closed with few interrupted absorbable sutures. The patient recovered well and was discharged on 11th postoperative day.

Drain site gut prolapse after abdominal surgery is a rare complication of drain insertion [2]. Increased morbidity and mortality have been noted in patients with drain site hernia, especially if strangulation of the loops of bowel sets in [3]. Other complications include drain site sepsis, bleeding from abdominal wall vessels, kinking and knotting of drains, and incisional hernia. Most reported cases of gut evisceration involved a drain site with an external diameter of greater than 10 mm. Predisposing factors for herniation through a drain site include general debility, increased intra-abdominal pressure and steroid administration [4]. The recommendations to reduce the risk of prolapse or hernia

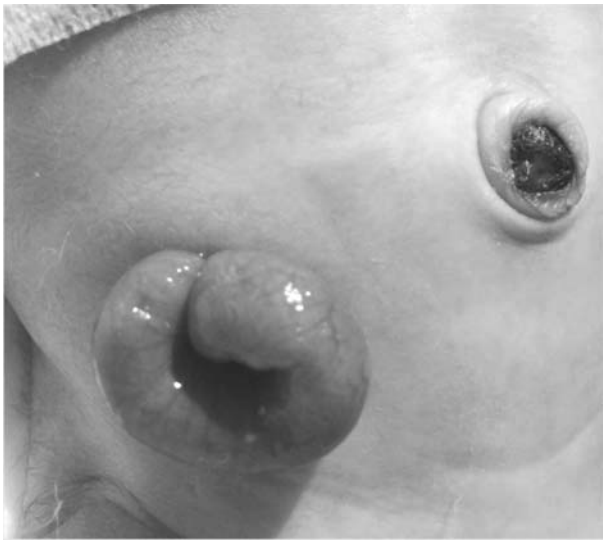

FIG.1 *Prolapse of gut through the drain site.*