DOI: https://doi.org/10.53350/pjmhs2023176529

ORIGINAL ARTICLE

Genetic and Developmental Factors to Consider in a Baby with Type 2 Ileal Atresia, Pectus Excavatum, Indirect Inguinal Hernia, and Bilateral Hydroceles

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ABSTRACT

Objective: The objective was to study a neonate who had type 2 ileal atresia, pectus excavatum, bilateral hydroceles, indirect inguinal hernia, and hereditary factors affecting development.

Methods: This observational study consisted of 135 neonates. Detailed demographic information of the cases that were enrolled was obtained after obtaining explicit written consent. Cases of pectus excavatum, bilateral water retention, type 2 ileal atresia, and indirect inguinal hernia are common. SPSS 22.0 was used to analyze all data.

Results: Mean gestational age of neonates was 38.12 weeks.Mean birth weight of neonate 3,027 g, length was 47.9cm, Family history of PE was found in 27 (20%) among all cases. Among all cases 37 (27.4%) had PE, bilateral hydroceles was found in 23 (17.03%) cases, indirect inguinal hernia in 19 (14.1%) and type 2 ileal atresia in 17 (12.6%).

Conclusion: This study's findings suggest a higher frequency of genetic syndromes in neonates and call for additional research into the causes of these anomalies, which may include developmental disruptions in structures derived from mesoderm or in utero vascular insults.

Keywords: Bilateral Hydroceles, Pectus Excavatum, Type 2 Ileal Atresia**,** Indirect Inguinal Hernia

INTRODUCTION

Although congenital defects often manifest independently, there may be common embryological, genetic, or environmental reasons in rare cases. An extremely unusual case of pectus excavatum (PE), bilateral water retention, and type 2 ileal atresia (IA) in a baby is detailed in this article. Approximately 95% of fetal chest wall abnormalities are caused by PE, which is also called "funnel chest"[1]. The xiphisternum is the most common site of the malformation, but it can be seen on the front of the chest, especially in the third to seventh costocartilages or ribs[1]. Present in around 1% to 2% of live births, PE is more common in male patients than female patients, with a ratio of men to women of 5:1[1].

Hydroceles occur when fluid accumulates within the tunica vaginalis of the testis [2]. In congenital hydrocele, the peritoneal cavity and scrotum are not linked because the processus vaginalis cannot close[2]. Hydroceles that communicate and those that do not are structurally different[2]. The difference between communicative and non-communicating hydroceles is that the former allows fluid to flow into the scrotum from the peritoneal cavity, while the latter does not[2]. Primary communicating hydroceles are the most common type in children; a vaginal patent is present in 80% to 90% of full-term male newborns[2]. The vast majority of instances include hydroceles on the right side of the body, although 25% of patients also have them on the left side, and 15% on both sides2]

There are two main forms of hernias: congenital and acquired. Inguinal hernias can be either direct or indirect. Indirect hernias happen when the intestines push past the inguinal rings and sometimes even into the scrotum, and this can happen to anyone who is born with a weak inguinal canal or who develops one over time[3]. Although they can never develop naturally, direct hernias are more common in men after middle age[3]. Despite not being part of the spermatic cord, they can nevertheless be found outside of it, within Hesselbach's triangle, medial to the lower epigastric arteries, and even extending into the scrotum via the inguinal canal[3]. One possible explanation for the preponderance of right-sided inguinal hernias (75% of cases) is the delayed descent and closure of the right testicle. Bilaterality affects 16% to 20% of children, with a third of those cases appearing before the baby is six months old[4]. Bilateral hernias affect approximately 10% of full-term infants and more than 50% of kids born prematurely or with a low birth weight[4]. Indirect hernias, which occur most often in juvenile patients, nearly always occur because the processus vaginalis stays open[5].

Intestinal adhesions impact between 5,000 and 14,000 live births annually, making them a prevalent cause of intestinal obstruction in newborns[6]. Multiple lesions or only one may be present; in most cases, the distal lesions don't show up until much later than the proximal ones[6]. Congenital deformities such as gastroschisis, cystic fibrosis, heart problems, and others can coexist with IA[6]. The fact that less than 10% of IA cases show extra-abdominal issues is probably due to the fact that vascular damage begins during pregnancy at a later stage[6]. While chromosomal abnormalities are found in fewer than 1% of instances, no clear link between IA and health difficulties in parents has been established[6].

Different types of intestinal atresia are categorized using the Grosfeld method, which is based on their distinct anatomical characteristics and frequency distributions. About one-fourth of cases of jejuno-ileal atresia are type 1, which does not have a mesenteric defect and has an internal membrane with serosal continuity[6,7]. Type 2, which comprises around 14% of cases[6,7], is characterized by a fibrous cord that connects the blind proximal and distal pouches with a serosal discontinuity. Type 3a, with a serosal discontinuity and a V-shaped mesenteric defect, accounts for approximately 16% of cases[6,7]. Type 3b, or "apple peel" atresia, is characterized by a small ileal segment curled around the ileocolic artery and affects about 9% of the population[6,7]. About 10% of cases are type 4, which is marked by several atresias[6,7]. Despite the fact that both conditions have been documented in the medical literature, the lack of a combined report raises concerns regarding potential syndromic or developmental connections. Results from an analysis of sixty-three cases with jejuno-ileal atresia were used to calculate the frequencies[6,7].

MATERIALS AND METHODS

This study was conducted at Department of Paediatric Surgery Qazi Hussain Ahmed Medical Complex Nowshera from December 2022 to May 2023 and comprised of 135 patients. After obtaining informed written consent detailed demographics of enrolled cases were recorded.

An X-ray of the abdomen was administered to each patient. The patient was assessed for bilateral hydroceles during the four-month follow-up appointment with the paediatric surgeon. The patient presented with a left-sided hydrocele that reached the cord, but there was no sign of a connection to the abdomen, according to the physical examination. On the right side, a suspected hernia

and a minor hydrocele were observed. The left side did not show any signs of inguinal hernias. In light of these results, the surgeon advised keeping a close eye on the patient and scheduling another evaluation in a year to look for any changes that would necessitate surgery. Any structural issues, such as the healing of the hydrocele or the emergence of a hernia, will be monitored throughout the long-term follow-up. To ensure complete care, it was necessary that pediatric surgery, pediatric gastroenterology, and general pediatrics work together, and that hospitals communicate effectively. We analyzed all of the data using SPSS 22.0. Cases of pectus excavatum, bilateral hydroceles, type 2 ileal atresia, and indirect inguinal hernia are common. Categorical variables were represented using percentages and frequencies.

RESULTS

Mean gestational age of neonates was 38.12 weeks. Mean birth weight of neonate 3,027.45 g, length was 47.9 cm. Family history of PE was found in 27 (20%) among all cases.(table 1)

Table-1: Baseline information of the neonates

Variables	Frequency (135)	Percentage
Mean gestational age (weeks)	38.12	
Mean birth weight (g)	3,027.45	
Mean length (cm)	47.9	
Family history of PE		
Yes	27	20
No	108	80

Among all cases 37 (27.4%) had PE, bilateral hydroceles was found in 23 (17.03%) cases, indirect inguinal hernia in 19 (14.1%) and type 2 ileal atresia in 17 (12.6%).(figure 1)

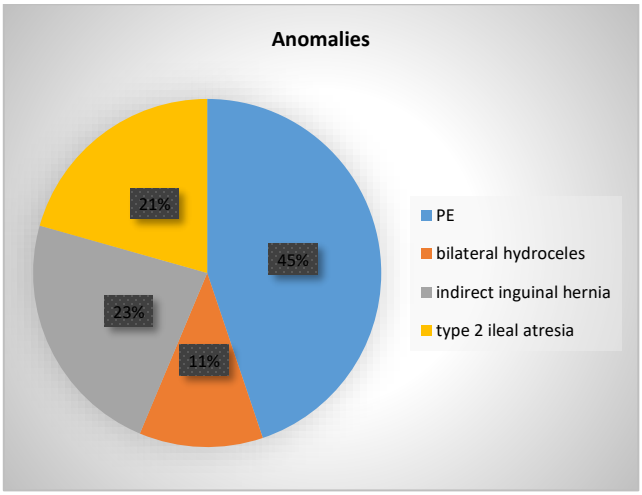

Figure-1: Frequency of anomalies among neonates

DISCUSSION

Among infants diagnosed with type 2 IA, bilateral hydroceles, indirect inguinal hernia, and PE, not a single case has been documented. This clinical picture is rare because all four illnesses occurred at the same time and there have been no analogous reports, even though individual occurrences of each sickness are common. Moreover, genetic testing may provide a shared explanation for this constellation of abnormalities. This may have its roots in the connective tissues, a shared progenitor in the germ cells, or even environmental factors that impact the developing baby.

Inguinal hernias, hydrocele, and petechiae are examples of congenital abnormalities that can result from issues with the mesodermal layer during development[8]. The fact that multiple conditions manifest in this patient at the same time raises the possibility that they have an underlying cause. Although these anomalies impact different anatomical systems, there have been reports of chest wall malformations occurring alongside genitourinary problems[9,10]. These symptoms do not have a prior record, however they are consistent with established syndromes such as Shprintzen-Goldberg, Aarskog-Scott, and Noonan syndromes[9]. While none of the above syndromes provide a full explanation for this patient's symptoms, they do demonstrate the existence of alternate syndromes that incorporate more than one of these features.

This patient has a history of embryology and also exhibits symptoms consistent with a connective tissue abnormality. Anomalies in extracellular matrix components, elastic fibers, and collagen are commonly linked to connective tissue illnesses, such as PE and hydrocele/inguinal hernias[9]. This individual exhibits symptoms of a communicating hydroceles on the right side and an indirect inguinal hernia on the right side[4]. A prolonged patent processus vaginalis causes congenital indirect hernias, as opposed to acquired direct inguinal hernias[8].

Notable connective tissue diseases including Marfan syndrome and Ehlers-Danlos syndrome (EDS) are associated with a range of systemic problems. Characteristic of Marfan syndrome, according to physical examination results, include PE and, less often, urogenital anomalies such as hydroceles[11]. Although there is no known causal relationship between type 2 IA and Marfan syndrome, both conditions are members of the same spectrum of connective tissue disorders, which can manifest in a variety of ways, including gastrointestinal issues[11]. In addition, PE is common in several subtypes of EDS. While there isn't much proof that EDS causes bilateral hydroceles or IA, the connective tissue laxity that is common in EDS could make hydroceles and other urogenital abnormalities worse[12].

The majority of congenital malformations, including type 2 intrauterine infections, bilateral hydroceles, and prematurity, are brought about by environmental factors, including inadequate prenatal nutrition, infections, and mechanical stresses. These factors influence embryonic development and raise the risk of anatomical anomalies when they interact with hereditary predisposition. Inadequate prenatal nutrition or metabolic abnormalities can impair vascular and lymphatic development, which in turn can impair drainage and lead to fluid buildup in the scrotal area, both of which can accelerate the development of hydrocele[13]. An increased incidence of some congenital malformations has also been associated with mothers' inadequate intake of certain minerals and vitamins, such as folic acid. The normal development of the baby's bones and cartilage depends on the mother's diet during pregnancy[13].

Abnormal foetal oxygen and nutrition delivery during pregnancy can disrupt the normal development of the gastrointestinal tract[13]. This is because changes to the uterine environment and a decline in placental function are caused by dietary deficiencies. The pathophysiology of intrauterine vascular accidents (IVAs) is thought to begin with ischemic necrosis of the fetal intestine, which affects the branches of the mesenteric arteries in the midgut[6]. There are a number of conditions that can disrupt the baby's blood vessels while they are still inside the mother's body, including intussusception, volvulus, and gastroschisis[6]. Intestinal atresia is more common in cases of thromboembolic occlusions, vasoconstrictive medications, and maternal smoking in the first trimester[6]. There is a lack of information regarding the mother's nutritional status and other environmental factors in the patient's medical record, which could have had a role in the detected anomalies.

Embryonic development can also be impacted by maternal illnesses, like intrauterine infections. These diseases can impact the maturation of the genitourinary system, which can lead to hydroceles[14]. Moreover, intrauterine variables such adhesions or constrictions may cause mechanical interference with the processes of intestinal development[6]. The increased risk of ischemia episodes generated by these mechanical and developmental abnormalities can also affect gut integrity during

critical phases of fetal development[6]. Intrauterine restriction, oligohydramnios, and adhesions are mechanical reasons of chest wall deformities that can happen during foetal development[15]. The growth and positioning of thoracic structures may be affected by mechanical variables[15-18]. Despite the lack of clarity regarding the underlying mechanical variables, it should not be disregarded that these aspects may potentially contribute to the reported anomalies.

The atretic segment must be surgically removed as soon as possible in order to prevent foetal intestinal ischemia necrosis, a medical emergency that is induced by IA pathogenesis[6]. In contrast, symptoms of conditions like hydrocele, inguinal hernia, and PE don't usually manifest until much later in life[1-4]. For instance, compared to the neonatal or infant periods, puberty is a more common time for cardiopulmonary issues associated with PE to appear or worsen[1]. Pulmonary function tests (PFTs), electrocardiograms (EKGs), and cardiac imaging are utilized to track the progression of PE[1]. Commonly treated with caution during infancy, hydroceles and inguinal hernias usually resolve themselves within a year[2-4]. In order to assess the need for additional intervention, this patient is booked for a follow-up session in one year.

We highlight the need for a comprehensive evaluation that includes genetic testing and propose that the patient's congenital abnormalities may be associated with another disorder. The identification of comorbidities that may impact the surgical or therapeutic plan could be facilitated by this type of assessment. For example, one study found that individuals with PE who also have a genetic connective tissue disorder should wait to have surgery to address their problem until their bones have fully developed[16-19].

CONCLUSION

This study's findings suggest a higher frequency of genetic syndromes in neonates and call for additional research into the causes of these anomalies, which may include developmental disruptions in structures derived from mesoderm or in utero vascular insults.

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