Case Report

Kartagener syndrome associated with esophageal atresia in a newborn. Clinical case.

A.Z. Kussainov[1,2], G.B. Altynbayeva[1,3], K.S. Ashirbay[1,3], E.B. Aitbayeva[4], G.T. Kaukenbayeva[1], G.S. Berdiyarova [1,3], M.M. Kalabayeva[1,3], N.B. Aflatonov[1,3], Y.V. Tyan[1], M.T. Duisebayev[1,2], Z.K. Tanat[5], Ye.B Kurakbayev[1], A.A. Rakhmatullaev[6]

ABSTRACT

Background

Kartagener syndrome is a rare form of primary ciliary dyskinesia, characterized by a triad of symptoms including situs inversus, chronic respiratory tract infections, and bronchiectasis. Esophageal atresia is a severe congenital anomaly requiring early surgical correction in the neonatal period. The combination of Kartagener syndrome with esophageal atresia is extremely rare and presents significant challenges for early diagnosis and management of newborns.

Objective

To present and analyze a clinical case of a newborn with a rare combination of Kartagener syndrome and esophageal atresia, paying special attention to the features of diagnosis, surgical treatment, and postoperative management in the context of complex combined congenital pathology.

Materials and Methods

The article presents a clinical case of a newborn with Kartagener syndrome associated with esophageal atresia. The features of the clinical picture, results of instrumental and laboratory diagnostic methods, as well as the surgical treatment strategy and postoperative management of the patient are described. Special attention is paid to diagnostic difficulties associated with atypical organ arrangement and the high risk of respiratory complications in the early neonatal period.

Results

In the presented clinical case of a newborn with a rare combination of Kartagener syndrome and esophageal atresia, the following results were obtained: Diagnostic stage: Thanks to a comprehensive preoperative examination (echocardiography, abdominal ultrasound, radiography), situs inversus with a right-sided aortic arch was timely diagnosed, which determined the choice of a left-sided surgical approach. Surgical treatment: Successful correction of esophageal atresia was performed through a left-sided thoracotomy: the tracheoesophageal fistula was closed, and a primary end-to-end esophagoesophagostomy was created. The early postoperative period was uneventful; anastomotic integrity was confirmed by control esophagography on the 9th day.

Postoperative complications and their management: On the 20th day, left-sided exudative pleurisy (lymphatic exudate) developed, requiring pleural cavity drainage. Combined therapy was administered: octreotide (10 mcg/kg for 14 days) and intrapleural administration of the drug "Lymphablock" (for 7 days), which led to adhesion formation and cessation of lymphorrhea.

Outcome: Enteral nutrition was restored on the 25th day after surgery. Compensated stenosis of the upper third of the esophagus was diagnosed, not requiring emergency intervention. The child was weaned to spontaneous breathing by the 45th day and discharged home on the 60th day of life in satisfactory condition for outpatient follow-up.

Key result: Timely topical diagnosis of organ arrangement anomalies and a multidisciplinary approach allowed for successful surgical correction and management of postoperative complications (chylothorax/lymphatic pleural effusion), despite the high respiratory risk associated with Kartagener syndrome.

Conclusion

In Kartagener syndrome, thorough echocardiography is necessary to clarify the location of the aortic arch in order to choose the correct surgical strategy (in this case, the side of approach). The clinical case of a newborn with Kartagener syndrome and esophageal atresia demonstrates the diagnostic and therapeutic challenges in the context of complex congenital pathology. Early detection and a multidisciplinary approach can improve the outcomes of surgical treatment and reduce the risks of postoperative complications.

Keywords

Kartagener syndrome; primary ciliary dyskinesia; esophageal atresia; newborn; congenital malformations; clinical case.

1. JSC "Scientific Center of Pediatrics and Pediatric Surgery", Almaty, Kazakhstan
2. NJSC "S.D. Asfendiyarov Kazakh National Medical University", Almaty, Kazakhstan
3. LLP "Kazakhstan Medical University 'Higher School of Public Health'", Almaty, Kazakhstan
4. NJSC "Al-Farabi Kazakh National University", Almaty, Kazakhstan
5. Astana Medical University, Pediatric surgery department
6. Tashkent State Medical University – Tashkent Medical Academy

Correspondence

G.B. Altynbayeva – PhD, Chief Physician, JSC "Scientific Center of Pediatrics and Pediatric Surgery"; Associate Professor, Head of the Department of Pediatrics, LLP "Kazakhstan Medical University 'Higher School of Public Health'", Almaty, Kazakhstan, e-mail: gulmira_altynbaeva@mail.ru,

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INTRODUCTION

Primary ciliary dyskinesia (PCD) is a rare (orphan) and genetically heterogeneous disorder characterized by structural and functional abnormalities of motile cilia in the respiratory epithelium. The clinical importance of PCD, particularly its most severe manifestation—Kartagener syndrome (defined by the triad of situs inversus, chronic sinusitis, and bronchiectasis)—is обусловлена not only the challenges associated with early and accurate diagnosis but also the progressive and multisystem nature of organ damage, which can lead to significant disability from early childhood[1,3,7].

Despite advancements in diagnostic technologies, verifying PCD remains a challenging task. This is due to the need for a combination of methods: nasal NO measurement, high-speed video microscopy analysis, transmission electron microscopy, and genetic testing. The implementation of the PCD Foundation consensus recommendations (2016) has systematized approaches to diagnosis and monitoring; however, issues of differential diagnosis with cystic fibrosis and other respiratory pathologies persist[1,2,8].

Cases with an atypical disease course and the identification of new genetic mutations present a particular challenge. For instance, recent studies demonstrate the role of CCNO gene mutations in the development of PCD in children, expanding the understanding of the disease's pathophysiology and necessitating the search for new therapeutic targets [6,7]. Furthermore, in adult patients, PCD is often diagnosed late, after irreversible bronchiectasis has already developed, highlighting the need for increased awareness among internists and pulmonologists [2,4].

The issue of male infertility in Kartagener syndrome, caused by immotile sperm flagella, deserves special attention. Understanding the genetic mechanisms underlying this process opens prospects for developing methods to overcome infertility and for medical genetic counseling [5].

The relevance of the topic is also heightened by the frequent association of PCD with other congenital anomalies, requiring a multidisciplinary approach to patient management. Cases combining situs inversus with surgical pathologies of the gastrointestinal tract are of particular interest. For example, the occurrence of duodenal atresia in patients with situs inversus necessitates thorough preoperative imaging to select the correct surgical approach[9,10]. Gastric rotation anomalies (dextrogastria), which can be either isolated or associated with diaphragmatic hernias, require antenatal diagnosis and timely correction to prevent life-threatening conditions [11-13].

Finally, the management of concomitant malformations (e.g., esophageal atresia with tracheoesophageal fistula) in such patients presents a significant surgical challenge. The debate regarding the choice of surgical method — thoracotomy or thoracoscopy — remains open. Contemporary research, including data from pediatric surgery consortia, compares the short-term and long-term outcomes of different approaches, analyzes risks associated with aortic arch anomalies, and explores the possibilities of endoscopic closure for recurrent fistulas[14-20]. This underscores the need for further accumulation of clinical experience and improvement of surgical techniques.

Thus, the issue of primary ciliary dyskinesia and its associated developmental anomalies is interdisciplinary, encompassing pediatrics, pulmonology, otorhinolaryngology, reproductive medicine, and surgery, which determines the high relevance of further research in this area.

Objective of the study: To present and analyze a clinical case of a newborn with a rare combination of Kartagener syndrome and esophageal atresia, paying special attention to the features of diagnosis, surgical treatment, and postoperative management in the context of complex combined congenital pathology.

Materials and Methods: The article presents a clinical case of a newborn with Kartagener syndrome associated with esophageal atresia. The features of the clinical picture, results of instrumental and laboratory diagnostic methods, as well as the surgical treatment strategy and postoperative management of the patient are described. Special attention is paid to diagnostic difficulties associated with atypical organ arrangement and the high risk of respiratory complications in the early neonatal period.

Patient Information: A 28-year-old woman, first pregnancy, first childbirth. The pregnancy was induced using the in vitro fertilization (IVF) method with embryo transfer. Prenatal ultrasound examination of the fetus, performed at 34 weeks of gestation, revealed a number of congenital malformations, including anomalies of the cardiovascular system: hypoplasia of the aorta and

suspected coarctation of the aorta. Esophageal atresia was diagnosed in the gastrointestinal tract. Significant polyhydramnios was noted. The mother was diagnosed with moderate anemia.

At 38 weeks of gestation, a full-term boy was born with a birth weight of 3106 g, length of 51 cm, and Apgar scores of 5 at the 1st minute and 6 at the 5th minute of life. The delivery was by planned cesarean section. After birth, the newborn presented with severe grade III respiratory failure, requiring tracheal intubation with a 3.5 mm tube and resuscitation measures in the delivery room.

Clinical Data: An attempt to insert a nasogastric tube revealed obstruction; the Elephant test was positive, confirming the suspicion of esophageal atresia. Initial mechanical ventilation parameters included FiO_2 32%, peak inspiratory pressure (PIP) 21 cm H_2O, positive end-expiratory pressure (PEEP) 5 cm H_2O, respiratory rate 40 breaths per minute, inspiratory time 0.45 seconds, and oxygen saturation (SpO_2) — 92%.

After stabilization of the child's condition, he was transferred from the Scientific Center of Obstetrics, Gynecology, and Perinatology to the specialized Neonatal Intensive Care Unit (NICU) of the Scientific Center of Pediatrics and Pediatric Surgery (Almaty, Kazakhstan).

Respiration was maintained on mechanical ventilation in PSIMV mode with the following parameters: inspiratory pressure (Pinsp) — 14 cm H_2O, pressure support (Psup) — 14 cm H_2O, PEEP — 7 cm H_2O, rate — 33 breaths per minute, inspiratory:expiratory ratio 1:2, FiO_2 — 60%. On auscultation, breath sounds were heard over all lung fields, and crepitant rales were detected. Heart sounds were heard on the right side, were muffled, with a regular rhythm.

Diagnostics: As part of the comprehensive examination, the following were performed:

1. Echocardiography — confirmed situs inversus, patent ductus arteriosus (2.2 mm) with insignificant left-to-right shunt, resistance index in the right pulmonary artery — 0.61, patent foramen ovale, dilation of the right heart chambers and the pulmonary artery trunk, normal left ventricular myocardial thickness and satisfactory systolic function, tricuspid and mitral regurgitation (+), systolic pulmonary artery pressure — 22 mm Hg, anomalous origin of the left coronary artery could not be excluded.
2. Neurosonography — revealed post-ischemic changes in the brain.
3. Ultrasound examination of the abdominal organs — visualized mirror-image arrangement of the organs (situs inversus).

Considering the echocardiography protocol, the right-sided location of the aortic arch, and the presence of Kartagener syndrome in the child, the strategy of left-sided thoracotomy was chosen.

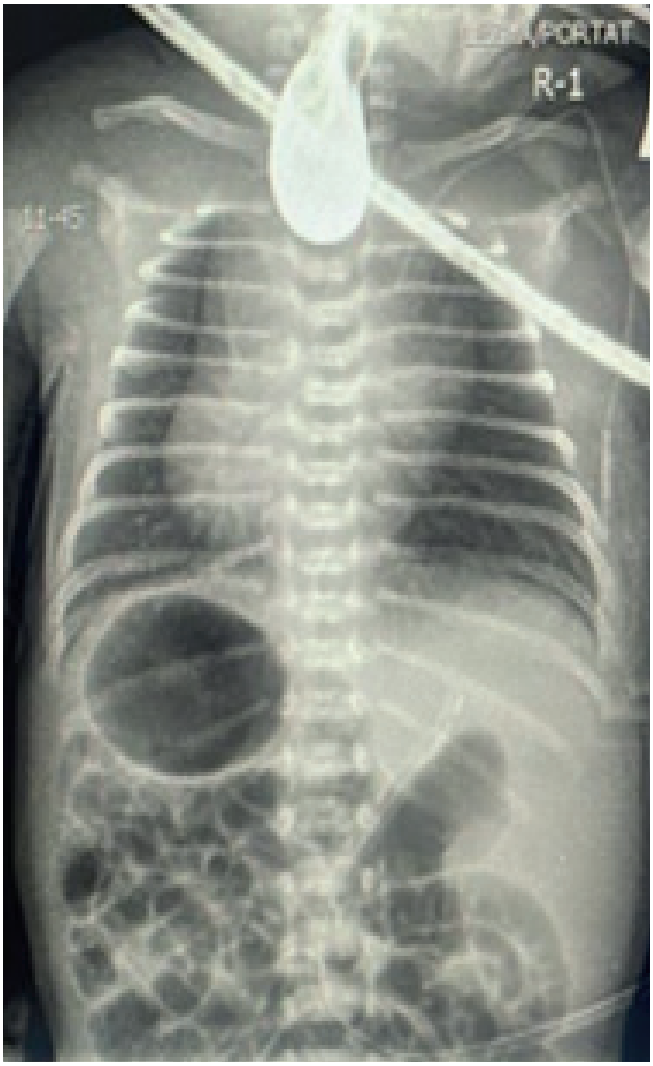

Figure 1 – Chest X-ray Survey Radiograph: Findings show esophageal atresia. Dextrocardia. Kartagener syndrome (situs inversus). Signs of right-sided pneumonia. Atelectasis of the right upper lobe. Fusion of the 9th and 10th ribs on the right.

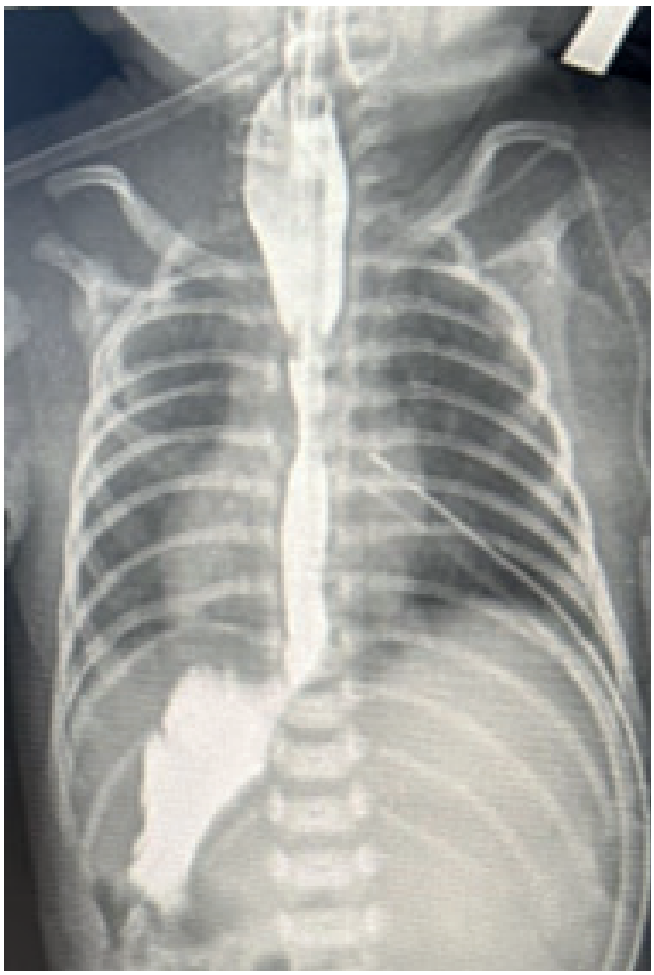

Figure 2 – 10-day-old Newborn. Diagnosis: Esophageal atresia with lower tracheoesophageal fistula. Dextrocardia. Kartagener syndrome (situs inversus). Postoperative condition after esophago-esophagostomy, anastomosis is intact.

Early postoperative period was uneventful. On the 9th day after surgery, a control esophagography was performed, which confirmed esophageal patency.

On the 10th day, the chest tube was removed due to the absence of fluid drainage from the pleural cavity. On the 11th day, the child was transitioned from mechanical ventilation to CPAP, and on the 15th day, transferred to a specialized department.

On the 20th day after surgery, progression of respiratory failure was noted; on auscultation, breath sounds were absent on the left side. Given the clinical findings, the child was transferred back to the Neonatal Intensive Care Unit (NICU), and a chest X-ray and ultrasound examination of the pleural cavity were performed.

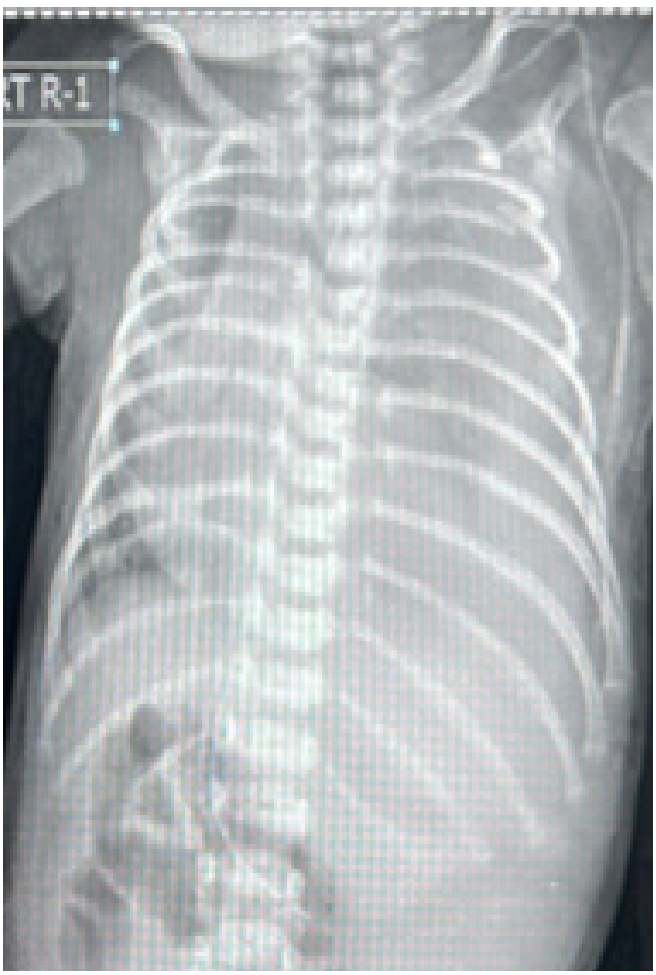

Figure 3 – Follow-up Chest X-ray: Fluid is observed in the left pleural cavity. The radiological conclusion revealed left-sided exudative pleurisy, and ultrasound confirmed a significant fluid accumulation in the left pleural cavity. Drainage was performed, yielding 90 ml of serous fluid. Bacteriological examination of the fluid indicated its lymphatic nature (chylous effusion/lymphatic exudate). Consequently, a decision was made to discontinue enteral nutrition and initiate therapy with octreotide at a dose of 10 mcg/kg.

Due to persistent fluid output (60–70 ml per day) despite octreotide therapy, the drug "Lymphablock" (10 ml volume) was administered into the left pleural cavity through the drainage tube once daily for 7 days. Follow-up ultrasound examination revealed the formation of multiple adhesions in the left hemithorax, which served as the basis for discontinuing Lymphablock.

Ethical Clearence

This clinical case was described in accordance with all ethical standards and principles provided by the Declaration of Helsinki. Informed consent was obtained from the patient's parents for publication and use of medical information for scientific analysis and dissemination. The patient's personal information was completely anonymized, excluding the possibility of identification. The study did not involve interventions beyond standard medical care and did not require separate approval from an ethics committee.

RESULTS

On the 25th day after surgery, enteral nutrition with a low-osmolarity formula was resumed. Octreotide was administered for a total of 14 days. Control fibroesophagogastroduodenoscopy revealed a compensated postoperative stenosis of the upper third of the esophagus. By the 45th day, the child was completely weaned from CPAP. On the 60th day of life, the child was discharged home with recommendations for further follow-up.

DISCUSSION

The presented clinical case represents a rare combination of Kartagener syndrome and esophageal atresia. The presence of situs inversus complicates diagnosis and surgical intervention, while functional insufficiency of the ciliated epithelium increases the risk of respiratory complications and complicates postoperative management. Treating esophageal atresia in newborns with concomitant primary ciliary dyskinesia requires an individualized approach involving a multidisciplinary team of specialists.

Comparison with the limited published data reveals both similarities and differences. In our case, the key diagnostic step determining the success of the operation was a thorough echocardiography to clarify the location of the aortic arch. Identifying a right-sided aortic arch justified the choice of left-sided thoracotomy, which aligns with the general surgical principle of selecting the approach in newborns with esophageal atresia and great vessel anomalies. The literature emphasizes that ignoring this step in patients with situs inversus can lead to fatal errors in surgical access [9,10,17].

In this case, timely diagnosis, adequate surgical treatment, and comprehensive postoperative management allowed for successfully managing complications, including the development of lymphatic pleurisy (chylothorax), which required medical therapy with octreotide and Lymphablock. Interestingly, the available literature on Kartagener syndrome primarily focuses on respiratory complications (pneumonia, bronchiectasis, sinusitis) [1,4,7,8], while specific surgical complications such as lymphatic pleurisy following esophageal atresia correction are practically undescribed in the context of PCD. This suggests that impaired ciliated epithelial function in Kartagener syndrome could potentially affect lymphatic clearance and slow down effusion resorption, although this hypothesis requires further investigation.

This case underscores the need for high clinical vigilance when managing newborns with complex congenital pathology and atypical organ arrangement, facilitating timely detection of complications and improving treatment efficacy. Accumulating such clinical observations is necessary for developing clearer management protocols for this extremely rare category of patients.

CONCLUSION

In Kartagener syndrome, thorough echocardiography is necessary to clarify the location of the aortic arch in order to choose the correct surgical strategy (in this case, the side of approach). The clinical case of a newborn with Kartagener syndrome and esophageal atresia demonstrates the diagnostic and therapeutic challenges in the context of complex congenital pathology. Early detection and a multidisciplinary approach can improve the outcomes of surgical treatment and reduce the risks of postoperative complications.

AUTHOR CONTRIBUTIONS

concept development – K.S. Ashirbay, Y.V. Tyan, G.S. Berdiyarova. study design – G.B. Altynbayeva, E.B. Aitbayeva. M.T. Duisebayev. conduct of the stated research – E.B. Aitbayeva, G.T. Kaukenbayeva. interpretation of the stated research – Z.K Tanat. M.M. Kalabayeva. manuscript preparation – A.Z. Kusainov. N.B. Aflatonov. E.B Kurakbayev

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REFERENCES

1. Shapiro A.J., Davis S.D., Polineni D., Manion M., Rosenfeld M., Dell S.D., Chilvers M.A., Ferkol T.W., Zariwala M.A., Sagel S.D., Josephson M., Morgan W., Yilmaz O., Olivier K.N., Milla C., Pittman J.E., Daniels M.L.A., Jones M.H., Marthin J.K., Schreck D.M., Loebinger M.R., Lucas J.S. Diagnosis, monitoring, and treatment of primary ciliary dyskinesia: PCD foundation consensus recommendations. *PLoS One*. 2016;**11**(12):e0152261.
2. Lucas J.S., Burgess A., Mitchison H.M., Moya E., Williamson M., Hogg C. Primary ciliary dyskinesia in adults: current diagnostic and management strategies. *Lancet Respir Med*. 2014;**2**(8):684-692.
3. Leigh M.W., Pittman J.E., Carson J.L., Ferkol T.W., Dell S.D., Davis S.D., Knowles M.R., Zariwala M.A. Clinical and genetic aspects of primary ciliary dyskinesia/Kartagener syndrome. *Genet Med*. 2009;**11**(7):473-487.
4. El Marzouki N., Alaoui-Inboui F., Slaoui B., Kamaoui I., Edderai M., Boumdin H. Kartagener's Syndrome: A Case Series. *Cureus*. 2024;**16**(6):e61722.
5. Sha Y.W., Ding L., Li P., Zhao Y. Management of primary ciliary dyskinesia/Kartagener's syndrome in infertile male patients and current progress in defining the underlying genetic mechanism. *Asian J Androl*. 2014;**16**(1):101-106.
6. Tong L., Li L., Wang W., Chen J., Li X., Zhang Y. Case Report: Primary ciliary dyskinesia due to CCNO mutations: a Chinese pediatric case series and literature review. *Front Pediatr*. 2024;12:1458660.
7. Blanchon S., Papon J.F., Beydon N., Nathan N., Thouvenin G., Bingen E., Fauroux B. Primary ciliary dyskinesia in children (Article in French). *J Pediatrie Pueric*. 2020;**33**:109-117.
8. Braun J.J., Boehm N., Metz-Favre C., Kessler R., de Blay F. Diagnosis of primary ciliary dyskinesia: when and how? (Article in French). *Ann Fr Oto-Rhino-Laryngol Pathol Cerv Fac*. 2017;**134**:360-365.
9. Tamina I.S. Kartagener Syndrome. Grodno Regional Children's Clinical Hospital. 22.04.2025. (Note: This source appears to be a local publication or report, with the author limited to the specified individual).
10. Qiang S., Fan M., Cui Q., Wang Y., Li S. Management of duodenal atresia associated with situs inversus abdominus: A case report. *Medicine*. 2020;**99**(31):e21439.
11. Saeki I., Yagi M., Nakayama M., Yamada W., Yamamoto M., Taguchi T. Transumbilical Surgery for Duodenal Stenosis in a Child with Situs Inversus: The First Report. *CORE. (Примечание: Полный состав авторов в доступных аннотациях может быть ограничен, указаны основные)*.
12. Versteegh H.P., Adams S.D., Stanton M.P., Burge D.M. Antenatally diagnosed right-sided stomach (dextrogastria): A rare rotational anomaly. *J Pediatr Surg*. 2016;**51**(2):236-239.
13. Aziz S., König S., Rasheed F.B., Zuberi S. Isolated dextrogastria with eventration of right hemidiaphragm and hiatal hernia in an adult male. *BMC Gastroenterol*. 2022;22(1):56.
14. Castro P., Riccetti S., Basta M., Ghirardo V., Gamba P., Midrio P. Association of Operative Approach With Postoperative Outcomes in Neonates Undergoing Surgical Repair of Esophageal Atresia and Tracheoesophageal Fistula. *J Pediatr Surg*. 2024;**59**(11):161641.
15. Marquart J.P., Bowder A.N., Bence C.M., Densmore J.C., Gourlay D.M., Arca M.J., Lal D.R., Sato T.T., Wagner A.J. (Midwest Pediatric Surgery Consortium). Thoracoscopy versus thoracotomy for esophageal atresia and tracheoesophageal fistula: Outcomes from the Midwest Pediatric Surgery Consortium. *J Pediatr Surg*. 2023;**58**(1):27-33.
16. Yalcin S., Bhatia A.M., He Z., Wulkan M.L., Durham M.M., Raval M.V., Pandya S.R., Shehata B.M. Short- and Long-Term Outcomes of Thoracoscopic and Open Repair for Esophageal Atresia and Tracheoesophageal Fistula. *J Pediatr Surg*. 2024;**59**(12):161662.
17. Yang Y.F., Dong R., Zheng C., Jin Z., Chen G., Huang Y., Zheng S. Outcomes of thoracoscopy versus thoracotomy for esophageal atresia with tracheoesophageal fistula repair: A PRISMA-compliant systematic review and meta-analysis. *Medicine*. 2016;**95**(30):e4428.
18. Ashirbay K, Dzhumabekov A, Kussainov A, Altynbayeva G, Aitbayeva E, Khairov K, et al. Aponeurotic Ring-Assisted Stoma Formation in Pediatric Anorectal Malformations: Surgical Stages and Early Outcomes. *Bangladesh Journal of Medical Science*. 2026;**25**(1):353-359. DOI: **https://doi.org/10.3329/bjms.v25i1.86436**
19. Ashirbay KS, Dzhumabekov AT, Kusainov AZ, Altynbayeva GB, Erekeshov AA, Aitbayeva EB, Kalabayeva MM, Kaukenbayeva GT, Shilanbaev NR, Zhaksylyk M. Evaluation of the effectiveness of intestinal stoma techniques in newborns with gastrointestinal tract defects. *Reproductive Medicine.* 2024;**1**:122-130. https://doi.org/10.37800/RM.1.2024.122-130
20. Ashirbay KS, Dzhumabekov AT, Kusainov AZ, Altynbayeva GB, Aitbayeva EB. Surgical design of stoma in children: A literary review. *Reproductive Medicine.* 2024;**1**:113-121. https://doi.org/10.37800/RM.1.2024.113-121