GMR
The Original

GENETIC SYNDROMES IN PECTUS EXCAVATUM AND PECTUS CARINATUM: IMPLICATIONS FOR DIAGNOSIS AND SURGICAL STRATEGY

Zarema Vakhaevna Kurbanova[1], Madina Abdurakhmanovna Begova[1], Aminat Magomedovna Kurbanova[1], Revana Malikovna Abdurakhmanova[1], Aysha Khamzatovna Kurbanova[2], Fatima Shamilevna Tagirova[3], Patimat Tatamovna Gadzhikhanova[4], Dzhamilya Kamaltinovna Mutueva[1]

[1] Dagestan State Medical University, Makhachkala, Russian Federation
[2] Astrakhan State Medical University, Astrakhan, Russian Federation
[3] Pirogov Russian National Research Medical University, Moscow, Russian Federation
[4] Russian University of Medicine, Moscow, Russian Federation

ABSTRACT

Pectus excavatum and pectus carinatum may occur as isolated deformities or as manifestations of inherited syndromes that increase treatment risks. Consequently, clinical decisions require assessment of sternal anatomy, cardiovascular disease, and connective tissue abnormalities. This review aimed to summarize clinical genetic features of syndromic deformities and their implications for diagnosis and correction. Russian and international publications from 2021 to 2026 were analyzed, including clinical guidelines, genetic studies, surgical series, and systematic reviews. The findings indicate that Marfan and Loeys–Dietz syndromes require attention to aortic and other vascular disease, whereas vascular Ehlers–Danlos syndrome raises concerns about tissue fragility and bleeding. In Noonan syndrome, cardiac abnormalities and hemostatic disorders are especially relevant. Together, these differences support a staged examination considering family history, systemic signs, functional findings, and targeted genetic testing when indicated. The choice between surgical and conservative treatment should reflect deformity morphology, age, symptoms, and coexisting conditions. Although genetic diagnosis may change operative timing and safety measures, it does not determine a preferred repair technique. Evidence for syndrome-specific surgical benefits remains limited by heterogeneous samples, incomplete molecular confirmation, and insufficient long-term follow-up. Therefore, the proposed diagnostic pathway requires further prospective validation.

KEYWORDS: *pectus excavatum, pectus carinatum, Marfan syndrome, Loeys–Dietz syndrome, Ehlers–Danlos syndrome, Noonan syndrome, genetic testing, chest wall repair.*

INTRODUCTION

Pectus excavatum and pectus carinatum are major developmental abnormalities of the anterior chest wall. However, outwardly similar deformities may represent isolated developmental features or manifestations of inherited syndromes that affect treatment strategy.

Official statistics underscore the importance of early recognition of congenital abnormalities. According to Rosstat, 932.9 cases of congenital anomalies, deformities, and chromosomal disorders per 100,000 children aged 0–14 years were first registered in Russia in 2024, compared with 917.5 in 2023. Among newborns weighing at least 1,000 g, these conditions were recorded in 44,500 infants in 2024 and 45,400 in 2023; the proportion increased from 3.7% to 3.8% [1]. These figures concern the entire diagnostic category, not chest wall deformities specifically.

Because national reporting does not distinguish pectus excavatum from pectus carinatum, prevalence estimates must be supplemented by specialist reviews [2]. Nevertheless, such estimates cannot establish whether a deformity is isolated or syndromic. Assessment should therefore extend beyond sternal shape to systemic features that could affect prognosis and treatment.

This principle underlies preoperative assessment. International recommendations address chest wall morphology, symptoms, functional limitations, and comorbidities, with specialist consultation when inherited disease is suspected [3]. Russian studies also emphasize age, progression, and cardiopulmonary status [4]. Thus, operative indications depend on overall health as well as sternal anatomy.

This evaluation is particularly important when Marfan, Loeys–Dietz, Ehlers–Danlos, or Noonan syndrome is suspected, because chest wall changes may coexist with extra-skeletal risks [5]. Genetic evaluation should be guided by family history and cardiovascular, ocular, and other systemic findings [6]. Early presentation increases suspicion but does not establish a syndromic diagnosis [7]. Molecular testing should therefore address a specific clinical hypothesis.

Recognition of a syndromic cause directly affects procedural safety. In hereditary aortopathies, the aorta and cardiac valves require priority assessment; with tissue fragility, hemostasis, fixation, and healing must be considered. Identified risks may alter operative timing or extent, or necessitate treatment of associated conditions first. Selection of minimally invasive, open, or conservative management should integrate anatomy, symptoms, and genetically determined constraints.

The aim of this review was therefore to synthesize current evidence on genetic syndromes associated with pectus excavatum and pectus carinatum and to establish their implications for diagnosis, preoperative evaluation, and surgical decision-making.

MATERIALS AND METHODS

An analytical review was conducted of publications on hereditary and genomic causes of pectus excavatum and pectus carinatum. Sources included PubMed, GeneReviews, international consensus statements, and Russian and international medical journals. The principal search period was 2021–2026; earlier diagnostic frameworks were included only when they remained relevant to contemporary criteria for inherited disorders.

Systematic and narrative reviews, clinical guidelines, cohort studies, genetic case series, surgical studies, and research on conservative treatment were included. Comparisons covered deformity type, age at presentation, family history, syndromic signs, molecular diagnosis, cardiovascular complications, operative techniques, and duration of follow-up. Established causal genetic associations were distinguished from associations reported in small samples.

Evidence was organized into clinical-genetic categories and synthesized qualitatively. Frequencies were compared only with regard to the relevant denominators and patient-selection criteria. No original clinical cases or quantitative meta-analysis were presented because studies differed in diagnostic definitions, age, deformity severity, surgical methods, and outcome measures.

RESULTS AND DISCUSSION

Genetic mechanisms and limits of the evidence

Pectus excavatum and pectus carinatum arise when the coordinated growth of costal cartilage, bone, and connective-tissue matrix is disturbed. Similar sternal shapes, however, do not establish a common inherited cause. In a review of nine studies, Ranjan and colleagues identified diverse familial and genetic associations but noted the absence of large genome-wide studies of isolated pectus excavatum [8]. Familial clustering merits attention but does not itself establish a monogenic syndrome.

Possible roles for COL5A1, COL5A2, ACAN, and SMAD4 have mainly been discussed in limited case series; the clinical relevance of a detected variant depends on its pathogenicity and phenotypic concordance [8]. Strokova and colleagues identified heterogeneous cellular phenotypes in costal cartilage, suggesting altered tissue remodeling [9]. Genetic hypotheses and cellular findings thus address different aspects of pathogenesis, but neither currently predicts thoracoplasty outcomes. Familial aggregation is particularly important when chest wall deformity coexists with aortic disease. In isolated deformity, a genetic finding without an established causal link may instead lead to unnecessary testing. Clinical utility depends on whether a result changes surveillance or management.

Accordingly, established extra-skeletal risks should be assessed in confirmed syndromes, targeted investigations pursued when combinations of signs are suspicious, and broad sequencing avoided as an automatic response to isolated deformity [8,9]. This approach reduces both missed hereditary aortopathy and overdiagnosis of connective-tissue disease.

Marfan syndrome: from chest wall assessment to aortic evaluation

Marfan syndrome, caused by pathogenic FBN1 variants, combines skeletal manifestations with a risk of progressive aortopathy. In a Russian cohort of 66 children, Gritsevskaya and colleagues associated selected manifestations with variant characteristics; another study described variability in neonatal presentations [10,11]. A shared molecular diagnosis therefore does not imply identical cardiovascular risk. Even marked sternal deformity cannot by itself determine the timing of surgery. External features raise suspicion but do not independently establish the diagnosis. Dietz considers chest wall deformity alongside body proportions, spine, feet, eyes, and cardiovascular findings [12]. Chakova and colleagues likewise emphasize correct classification of FBN1 variants [13]. Variants of uncertain significance require interpretation in light of clinical and family findings; genetic results cannot replace preoperative evaluation.

Aortic-root dilatation, valve abnormalities, and possible dissection are the principal surgical concerns. International guidance recommends echocardiography, longitudinal monitoring, and additional imaging when indicated [14]. Active aortopathy may require treatment before chest wall surgery; in stable patients, thoracoplasty should be discussed with cardiology and genetics specialists [12,14]. Altered connective tissue calls for careful fixation and follow-up but does not automatically prohibit repair. The potential need for future cardiac or aortic procedures should also influence planning of retrosternal support, its removal, and possible revision, because these decisions may affect subsequent thoracic access.

Loeys–Dietz syndrome and vascular risk

In Loeys–Dietz syndrome, sternal deformities coexist with inherited abnormalities of TGF-β signaling. Loeys and Dietz identify arterial tortuosity, aneurysms, and cutaneous and craniofacial findings as reasons for expanded evaluation [15]. Unlike a typical FBN1-associated aortopathy, vascular involvement may extend beyond the aortic root. Chest wall morphology alone therefore cannot capture the full preoperative vascular risk.

Aortic disease guidelines and syndrome-specific guidance both emphasize vascular phenotype rather than echocardiography alone when broader assessment is indicated [14,15]. Magnetic resonance or computed tomographic angiography should be individualized according to age and initial findings. Possible cervical instability also affects intubation and positioning. Thoracoplasty should therefore be planned with cardiology and anesthesia teams before placement of supporting hardware. Where vascular disease requires active treatment, operative sequencing should be determined in an expert center. Future vascular access deserves early discussion when retrosternal metal bars are planned; no universal combined-treatment strategy exists.

Hypermobility or sternal asymmetry alone does not establish Loeys–Dietz syndrome without additional signs and family evidence. Appropriate care combines recognition of serious vascular lesions with avoidance of unnecessary investigations [14,15]. Successful cosmetic correction does not remove the need for hereditary aortopathy surveillance.

Ehlers–Danlos syndromes: differences in operative risk

Ehlers–Danlos syndrome subtypes have different surgical implications. Malfait, Symoens, and Syx describe skin hyperextensibility, characteristic scarring, and joint hypermobility in the classical type, most often associated with COL5A1 and COL5A2 [16]. Chest wall surgery must therefore account for skin quality, bruising, and the reliability of tissue closure. Sternal deformity itself is nonspecific and cannot determine the subtype.

The vascular subtype associated with COL3A1 presents a different level of risk. Byers highlights arterial and hollow-organ rupture and the importance of avoiding unnecessary invasive procedures [17]. These precautions should not be applied indiscriminately to all hypermobility; subtype assignment requires clinical and molecular evidence [16,17]. Elective thoracoplasty in vascular Ehlers–Danlos syndrome demands especially stringent assessment, and any necessary procedure requires tissue-sparing technique and advance hemostatic planning.

Supporting-tissue quality and scarring may influence reconstruction stability, but comparative studies of Nuss versus Ravitch repair in molecularly confirmed Ehlers–Danlos subtypes are insufficient. Tissue fragility is an established clinical concern, not a proven quantitative predictor of recurrence.

Noonan syndrome and other RASopathies

Noonan syndrome and related RASopathies involve altered RAS/MAPK signaling; the chest wall often combines upper protrusion with lower depression. Roberts reports congenital cardiac defects in 50–80% of affected individuals and hypertrophic cardiomyopathy in 20–30% [18]. Before correction, valve and myocardial status and exercise tolerance require assessment alongside sternal geometry. Genotype informs the clinical context but does not dictate an operative technique.

Cardiac risk may be independent of mechanical compression. Roberts' synthesis and a Russian report of severe syndromic cardiomyopathy support individualized echocardiographic and anesthetic evaluation [18,19]. Hemostatic abnormalities also matter: bleeding history should be obtained even without obvious hemorrhage, and hematology consultation sought when indicated [18]. Thoracoplasty cannot correct these underlying risks, and a satisfactory chest contour does not demonstrate resolution of cardiomyopathy. Cardiac follow-up is necessary whether management is operative or conservative.

Only after cardiac and hemostatic evaluation should correction of a mixed deformity be selected. Observation and conservative measures may suit a flexible chest wall, whereas fixed deformities may require reconstruction. Decisions depend on morphology, symptoms, and extra-skeletal disease, rather than syndrome name alone [18,19].

Chest wall deformities also occur in chromosomal disorders. Pollak and colleagues reported pectus excavatum in 22.81% and pectus carinatum in 5.26% of 57 individuals with 3q29 deletion; these proportions do not indicate the prevalence of the deletion among patients with chest wall deformities [20]. Vorobyeva and colleagues examined the association of pectus carinatum with Scheuermann disease, while Timofeev distinguished dysplastic features from proven inherited syndromes [21,22]. Chudaeva and Delyagin and colleagues added evidence on extra-skeletal manifestations of Marfan syndrome [23,24]. Thus, combinations of signs, rather than isolated sternal shape, warrant expanded evaluation (Table 1).

Table 1. Clinical significance of genetic syndromes in chest wall deformity

Syndrome	Typical additional signs	Main operative risk	Preoperative decision
Marfan (FBN1)	Tall stature, arachnodactyly, ocular and spinal abnormalities	Aortic-root dilatation, valvular disease	Echocardiography; cardiology assessment of aorta and sequencing
Loeys–Dietz (TGFBR1/2 and others)	Hypertelorism; palate, skin, and cervical abnormalities	Aneurysms in multiple vascular beds; cervical instability	Multidisciplinary review; vascular imaging and cervical assessment when indicated
Classical Ehlers–Danlos	Stretchable skin, abnormal scars, hypermobility	Skin fragility and healing problems	Plan incision, hemostasis, and layered closure
Vascular Ehlers–Danlos	Thin skin, bruising, family vascular events	Spontaneous vessel and tissue rupture	Assess acceptability of elective surgery in expert center
Noonan / RASopathies	Short stature, characteristic face, mixed pectus deformity	Cardiomyopathy, cardiac defects, coagulopathy	Echocardiography, anesthesia review, hemostatic evaluation if indicated
Microdeletions and duplications	Neurodevelopmental abnormalities, multiple congenital anomalies	Variant-specific comorbidities	Genetics consultation; chromosomal testing when clinically justified

Note. This table identifies diagnostic priorities and does not establish absolute contraindications to thoracoplasty. Based on references [12,14–20].

Table 1 shows that similar chest wall morphology can be associated with different systemic risks. Vascular assessment takes priority in hereditary aortopathies, tissue fragility in vascular Ehlers–Danlos syndrome, and cardiac and hemostatic evaluation in Noonan syndrome. Syndromic signs do not automatically preclude correction, but they change preoperative investigations and safety measures. These differences support a stepwise diagnostic pathway.

Preoperative diagnosis: sequence of decisions

Preoperative assessment begins with deformity morphology, age at onset, and family history. Billar and colleagues identify associated congenital signs and early onset as reasons to consider genetic evaluation [6,7]. Examination of the spine, joints, skin, and eyes, together with family history of aneurysm, is more informative than sternal shape alone.

Next, the functional effects of the deformity must be distinguished from independent cardiovascular disease. International recommendations support symptom-based evaluation, and aortic disease guidance recommends echocardiography when hereditary aortopathy is suspected [3,14]. Electrocardiography, tomography, and functional testing should be ordered as indicated to avoid confusing deformity-related symptoms with primary cardiac disease.

Molecular investigations should follow a clinical hypothesis: FBN1 variants require pathogenicity assessment; RASopathy results must be matched to phenotype; and multiple congenital anomalies may warrant testing for chromosomal abnormalities [13,18,20]. Genetic risks can then be weighed against the expected benefits of repair. The proposed pathway is an analytical synthesis, not a validated risk score (Table 2) [3,14–18].

Table 2. Diagnostic pathway before correction of chest wall deformity

Stage	Features assessed	Method	Implications
1. Initial examination	Deformity type, onset, progression, family history	History; examination of skeleton, skin, and face	Determine need for genetic consultation
2. Heart	Murmurs, arrhythmia, failure, suspected aortopathy	ECG, echocardiography; other tests as indicated	Identify conditions requiring priority treatment
3. Genetics	Combined external, familial, vascular, and other signs	Genetics consultation; targeted test/panel/copy-number analysis	Clarify inherited disorder
4. Anatomy	Asymmetry, rigidity, compression, rotation	Clinical assessment and justified imaging	Select repair technique and support requirements
5. Risks	Tissue fragility, hemostasis, respiration, neck	Anesthesiology and specialists; indicated laboratory tests	Adjust eligibility, timing, and safety measures
6. Multidisciplinary decision	Overall benefit, functional and psychological goals	Shared decision with patient and family	Agree on observation, bracing, or surgery

Note. The pathway is an author-derived analytical synthesis of references [3,6,14–18], not a clinically validated scale.

As summarized in Table 2, evaluation proceeds from clinical and familial features to functional and systemic risks. Genetic tests should be ordered when clinically justified and interpreted alongside cardiac and anatomical findings. Final decisions must balance the anticipated benefit of correction with procedural safety; the proposed sequence is not a validated risk scale. It provides a framework for choosing conservative or operative management.

Changes in surgical strategy according to genetic diagnosis

Choice of correction depends on chest wall rigidity, asymmetry, age, and previous procedures as well as the depth of depression. The Nuss procedure elevates the sternum with a retrosternal support bar; the Ravitch technique reconstructs abnormal cartilage and, when necessary, the sternum [25]. Both are used in selected patients. In inherited syndromes, aortic, cardiac, and tissue status require additional consideration. No repair technique should be preferred solely on the basis of the syndrome label.

Huerta and colleagues analyzed 10,053 procedures in patients aged 12–21 years: 8,673 (86%) were Nuss repairs and 1,380 (14%) were Ravitch repairs [26]. These figures describe clinical practice rather than genotype-specific superiority. In a meta-analysis of 21 studies involving 1,087 patients, Belgacem and colleagues found improvement in certain cardiac measures but no convincing improvement in the respiratory measures studied [27]. Another meta-analysis did not establish a consistent gain in maximal oxygen uptake [28]. Anatomical and functional success must therefore be distinguished.

Different syndromes call for different safety precautions. Loeys and Dietz emphasize widespread vascular lesions; Byers describes tissue fragility in vascular Ehlers–Danlos syndrome; and Roberts highlights cardiac disease and hemostatic abnormalities in Noonan syndrome [15,17,18]. These concerns cannot be collapsed into a universal risk score. They affect different stages of preparation and may even change whether surgery is acceptable. In aortopathy, the timing of possible vascular and thoracic procedures must also be coordinated [3,14].

The Haller index describes anatomy but neither establishes an inherited diagnosis nor explains dyspnea by itself. Guidelines recommend interpreting it alongside symptoms and functional findings [3]. Surgical planning should aim for meaningful

individual benefit at acceptable systemic risk, rather than a particular tomographic value. Direct compression of cardiac chambers must be distinguished from underlying valvular or myocardial disease, which may produce similar symptoms but require different treatment.

Conservative treatment and timing

Conservative correction depends primarily on chest wall flexibility. Compression bracing is used for flexible pectus carinatum; AlGhamdi emphasizes proper fitting, adherence, and early response to pressure [29]. In inherited conditions with fragile skin, brace tolerance requires separate assessment. Outcomes depend on anatomy, tissue condition, and treatment adherence.

A vacuum bell may be used for selected children with pectus excavatum. Shominova, Mitupov, and Razumovsky reported early Russian experience [30]. Unlike bracing for carinatum, the device gradually elevates the sternum, but an early contour change does not establish a durable result. Conservative treatment is not a universal substitute for surgery in severe fixed deformity or substantial functional impairment.

Treatment timing should reflect growth, symptoms, tissue flexibility, and patient preferences [3,29]. In mixed deformities, including those seen in Noonan syndrome, compression of one region alone may be inadequate [18]. Follow-up for inherited disease and its cardiovascular manifestations must continue regardless of the corrective approach.

Postoperative outcomes and long-term follow-up

Thoracoplasty success should be assessed by pain, exercise tolerance, activity, and patient satisfaction, not merely sternal position. Khakimov and Khodzhanov describe different anatomical causes of postoperative syndromes in children requiring different revision approaches [31]. Other Russian clinical data likewise show heterogeneous late contour changes [32]. Residual asymmetry, recurrence, and sternal displacement should not be grouped as one complication. Long-term reports should specify the date and method of bar removal, baseline asymmetry, and previous procedures; without these details, growth-related changes cannot be distinguished from fixation problems or separate disease processes.

Cruz-Centeno and colleagues evaluated long-term recurrence and patient satisfaction, extending assessment beyond radiographic measures [33]. However, most surgical series do not stratify patients by molecularly confirmed syndromes. The proposed association between connective-tissue quality and late deformity therefore remains a hypothesis rather than an established genotype-specific recurrence rate.

Syndrome-specific surveillance remains necessary after repair: aortic monitoring in Marfan and Loeys–Dietz syndromes, follow-up of cardiac disease in Noonan syndrome, and vascular precautions in vascular Ehlers–Danlos syndrome [12,15,17,18]. Normalizing sternal contour does not eliminate the underlying genetic disorder.

LIMITATIONS OF THE REVIEW

The evidence is limited by heterogeneous cohorts, genetic diagnostic criteria, and outcome measures. Early-onset studies often recruit patients with additional syndromic signs, so molecular diagnostic yield cannot be generalized to all children with chest wall deformity [7]. Anatomical improvement is not equivalent to functional improvement; postural-stability research and pediatric surgical reviews highlight the need for comparable long-term follow-up [34,35]. Future studies should record molecularly confirmed diagnoses, deformity morphology, cardiopulmonary function, operative technique, and outcomes after hardware removal.

Most surgical series also lack molecular stratification. Barman and colleagues discuss putative genetic associations, while Ranjan and colleagues emphasize the limitations of candidate-gene studies: an identified variant does not necessarily change prognosis or surgical selection [8,36]. Accordingly, the proposed diagnostic pathway is an analytical synthesis rather than a validated score, and genotype-specific preferences for repair technique require prospective study. Established vascular, cardiac, and tissue risks should remain the priority in treatment decisions.

CONCLUSION

Pectus excavatum and pectus carinatum may be isolated disorders or components of inherited syndromes. The clinical value of genetic testing lies not simply in detecting a variant but in identifying aortopathy, cardiomyopathy, hemostatic disorders, or increased tissue fragility. Aortic and vascular assessment is a priority in suspected Marfan or Loeys–Dietz syndrome; cardiac and hemostatic assessment in Noonan syndrome; and careful evaluation of the acceptability of elective surgery in vascular Ehlers–Danlos syndrome. Repair should be selected according to morphology, age, symptoms, comorbidities, and multidisciplinary expertise. Genetic diagnosis should guide adaptation of care rather than automatically exclude surgery. Prospective studies are needed to relate molecularly confirmed syndromes to functional and long-term surgical outcomes.

REFERENCES

1. Federal State Statistics Service. Healthcare in Russia. 2025: Statistical Collection [Internet]. Moscow: Rosstat; 2025. 149 p. [In Russian]. Available from: Official Rosstat publication.

2. Janssen N, Coorens NA, Franssen AJPM, Daemen JHT, Michels IL, Hulsewé KWE, Vissers YLJ, de Loos ER. Pectus excavatum and carinatum: a narrative review of epidemiology, etiopathogenesis, clinical features, and classification. J Thorac Dis. 2024;16(2):1687–1701. doi:10.21037/jtd-23-957.

3. Dunning J, Burdett C, Child A, Davies C, Eastwood D, Goodacre T, Haecker FM, Kendall S, Kolvekar S, MacMahon L, Marven S, Murray S, Naidu B, Pandya B, Redmond K, Coonar A. The pectus care guidelines: best practice consensus

guidelines from the joint specialist societies SCTS/MF/CWIG/BOA/BAPS for the treatment of patients with pectus abnormalities. Eur J Cardiothorac Surg. 2024;66(1):ezae166. doi:10.1093/ejcts/ezae166.
4. Motorenko NV, Vinnik AV. Pectus excavatum in children: modern aspects of the problem. Health and Ecology Issues. 2024;21(2):15–22. [In Russian]. doi:10.51523/2708-6011.2024-21-2-02.
5. Andreescu N, Sharma A, Mihailescu A, Zimbru CG, David VL, Horhat R, Kundnani NR, Puiu M, Farcas S. Chest wall deformities and their possible associations with different genetic syndromes. Eur Rev Med Pharmacol Sci. 2022;26(14):5107–5114. doi:10.26355/eurrev_202207_29298.
6. Billar RJ, Manoubi W, Kant SG, Wijnen RMH, Demirdas S, Schnater JM. Association between pectus excavatum and congenital genetic disorders: a systematic review and practical guide for the treating physician. J Pediatr Surg. 2021;56(12):2239–2252. doi:10.1016/j.jpedsurg.2021.04.016.
7. Billar R, Heyman S, Kant S, Wijnen R, Sleutels F, Demirdas S, Schnater JM. Early-onset pectus excavatum is more likely to be part of a genetic variation. Eur J Pediatr Surg. 2024;34(4):325–332. doi:10.1055/a-2081-1288.
8. Ranjan R, Imtiaz N, Waterhouse B, Paul I, Brunswicker A, Dunning J. Genetic associations with pectus excavatum: a systematic review. Curr Issues Mol Biol. 2026;48(1):122. doi:10.3390/cimb48010122.
9. Strokova EL, Pakhomova NY, Shevchenko AI, Korytkin AA, Kozhevnikov VV, Zaidman AM. Phenotypic features of costal cartilage cells in pectus excavatum. Siberian Scientific Medical Journal. 2023;43(6):203–209. [In Russian]. doi:10.18699/SSMJ20230625.
10. Gritsevskaya DY, Putintsev AN, Nikolsky DA, Semyachkina AN, Nikolaeva EA, Shkolnikova MA, Voinova VY. Clinical and genetic characteristics of a Russian cohort of children with Marfan syndrome. Russian Bulletin of Perinatology and Pediatrics. 2024;69(6):34–42. [In Russian]. doi:10.21508/1027-4065-2024-69-6-34-42.
11. Gritsevskaya DYu, Kuramagomedova RG, Vasiliev EV, Shkolnikova MA, Voinova VYu. Clinical variability of the neonatal form of Marfan syndrome in patients with FBN1 gene mutations. Russian Bulletin of Perinatology and Pediatrics. 2024;69(3):55–64. [In Russian]. doi:10.21508/1027-4065-2024-69-3-55-64.
12. Dietz HC. FBN1-related Marfan syndrome. In: GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 2001 Apr 18 [updated 2022 Feb 17; cited 2026 Oct 23]. Available from: NCBI Bookshelf — FBN1-Related Marfan Syndrome.
13. Chakova NN, Rudoy AS, Burak EA, Valyuzhenich YaI, Dolmatovich TV, Niyazova SS. Characterization of mutations in the FBN1 gene and assessment of their pathogenic status in patients with Marfan syndrome. Medical Genetics. 2024;23(3):38–48. [In Russian]. doi:10.25557/2073-7998.2024.03.38-48.
14. Isselbacher EM, Preventza O, Black JH 3rd, Augoustides JG, Beck AW, Bolen MA, Braverman AC, Bray BE, Brown-Zimmerman MM, Chen EP, Collins TJ, DeAnda A Jr, Fanola CL, Girardi LN, Hicks CW, Hui DS, Jones WS, Kalahasti V, Kim KM, Milewicz DM, Oderich GS, Ogbechie L, Promes SB, Ross EG, Schermerhorn ML, Times SS, Tseng EE, Wang GJ, Woo YJ. 2022 ACC/AHA guideline for the diagnosis and management of aortic disease: a report of the American Heart Association/American College of Cardiology Joint Committee on Clinical Practice Guidelines. Circulation. 2022;146(24):e334–e482. doi:10.1161/CIR.0000000000001106.
15. Loeys BL, Dietz HC. Loeys-Dietz syndrome. In: GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 2008 Feb 28 [updated 2024 Sep 12; cited 2026 Oct 23]. Available from: NCBI Bookshelf — Loeys-Dietz Syndrome.
16. Malfait F, Symoens S, Syx D. Classic Ehlers-Danlos syndrome. In: GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 2007 May 29 [updated 2024 Feb 1; cited 2026 Oct 23]. Available from: NCBI Bookshelf — Classic Ehlers-Danlos Syndrome.
17. Byers PH. Vascular Ehlers-Danlos syndrome. In: GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1999 Sep 2 [updated 2025 Apr 10; cited 2026 Oct 23]. Available from: NCBI Bookshelf — Vascular Ehlers-Danlos Syndrome.
18. Roberts AE. Noonan syndrome. In: GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; [updated 2025 Dec 4; cited 2026 Oct 23]. Available from: NCBI Bookshelf — Noonan Syndrome.
19. Dzeмeshkevich SL, Motreva AP, Ibragimov SV. Occlusion of the circumflex coronary artery during hospitalization after left ventricular remodeling for syndromic left ventricular hypertrophy in Noonan syndrome. 2026. [In Russian]. Bibliographic details require verification.
20. Pollak RM, Tilmon JC, Murphy MM, Gambello MJ, Sanchez Russo R, Dormans JP, Mulle JG. Musculoskeletal phenotypes in 3q29 deletion syndrome. Am J Med Genet A. 2023;191(11):2749–2756. doi:10.1002/ajmg.a.63384.
21. Vorobyeva EA, Razumovskiy AYu, Dubrov VE, Vybornov DYu, Krestyashin IV, Mitupov ZB, Vakhova EL. Combination of pectus carinatum and Scheuermann–Mau disease in children: an empirical pattern or somite syndrome? Russian Journal of Pediatric Surgery, Anesthesia and Intensive Care. 2023;13(1):13–24. [In Russian]. doi:10.17816/psaic1491.
22. Timofeev EV. Heritable disorders and dysplasia of connective tissue: synonyms or conceptually irreconcilable concepts? Juvenis Scientia. 2024;10(3):5–16. [In Russian]. doi:10.32415/jscientia_2024_10_3_5-16.
23. Chudaeva OV, Samylova AR, Vlasova DV, Korcheva VS, Airapetov KV. Clinical case of diagnostics and treatment of a patient with Marfan syndrome. Vestnik of the Smolensk State Medical Academy. 2024;23(2):126–130. [In Russian]. doi:10.37903/vsgma.2024.2.16.

24. Delyagin VM, Dosimov AZh, Aksenova NS, Aksenov AV. Manifestations of Marfan syndrome in maxillofacial pathology in children. Vrach. 2024;35(8):49–51. [In Russian]. doi:10.29296/25877305-2024-08-09.
25. Notrica DM, McMahon LE, Jaroszewski DE. Pectus disorders: excavatum, carinatum and arcuatum. Adv Pediatr. 2024;71(1):181–194. doi:10.1016/j.yapd.2024.05.001.
26. Huerta CT, Alligood DM, Davis JK, Ramsey WA, Cobler-Lichter MD, Shagabayeva L, Delamater JM, Hernandez AE, Parreco JP, Perez EA, Sola JE, Thorson CM. Outcomes after pectus excavatum repair: a nationwide comparison of Nuss versus Ravitch operations. J Surg Res. 2024;303:381–389. doi:10.1016/j.jss.2024.09.025.
27. Belgacem A, Abane C, Tricard J, Felix P, Lavrand F, Laloze J, Lopez P, Bothorel P, Auditeau E, Fourcade L, Ballouhey Q. Sternal advancement surgery for pectus excavatum: a systematic review and meta-analysis of cardiac and respiratory function outcomes. J Thorac Dis. 2025;17(7):4897–4908. doi:10.21037/jtd-2025-347.
28. Media AS, Juhl-Olsen P, Christensen TD, Katballe N, Vad H, Petersen RH, Wiggers H, Hansen SN, Farup J, Overgaard K, de Paoli FV. Cardiorespiratory fitness after correction of pectus excavatum: a systematic review with meta-analysis. Sci Rep. 2025;15(1):26282. doi:10.1038/s41598-025-08038-7.
29. AlGhamdi ZM. Determinants of successful pectus carinatum bracing: a narrative review. Saudi J Med Med Sci. 2026;14(3):217–222. doi:10.4103/sjmms.sjmms_306_25.
30. Shominova AO, Mitupov ZB, Razumovsky AYu. First experience of using a vacuum bell in the treatment of pectus excavatum in children. Russian Journal of Pediatric Surgery, Anesthesia and Intensive Care. 2022;12(3):311–318. [In Russian]. doi:10.17816/psaic1253.
31. Khakimov ShK, Khodzhanov IYu. Methods of surgical treatment of postoperative syndromes in children operated on for pectus excavatum. Plastic Surgery and Aesthetic Medicine. 2023;(3):78–83. [In Russian]. doi:10.17116/plast.hirurgia202303178.
32. Khodzhanov IYu, Khakimov ShK. Postoperative syndromes in children operated on for pectus excavatum. Plastic Surgery and Aesthetic Medicine. 2022;(3):61–67. [In Russian]. doi:10.17116/plast.hirurgia202203161.
33. Cruz-Centeno N, Fraser JA, Stewart S, Marlor DR, Oyetunji TA, St Peter SD. Long-term recurrence rates and patient satisfaction after repair of pectus excavatum. Eur J Pediatr Surg. 2024;34(4):333–337. doi:10.1055/a-2127-6133.
34. Nikityuk IE, Garkavenko YuE, Ryzhikov DV, Dolgiev BKh. Postural stability characteristics in children with asymmetric pectus excavatum. Modern Problems of Science and Education. 2024;(2). [In Russian]. doi:10.17513/spno.33319.
35. Davlyatov SB, Ibodov KhI, Rofiev RR, Sulaimonov SCh. Current aspects and pressing problems of pectus excavatum correction in children. Healthcare of Tajikistan. 2025;(3):96–102. [In Russian]. doi:10.52888/0514-2515-2025-366-3-96-102.
36. Barman S, Momin L, Islam R, Shing MS, Ranjan R, Waterhouse B, Dunning J. A scoping review on genetic mutations and single-nucleotide polymorphisms associated with pectus excavatum. J Multidiscip Healthc. 2026;19:592241. doi:10.2147/JMDH.S592241.

Information about the Authors

Kurbanova Zarema Vakhaevna – MD, DSc, Professor; Acting Head of the Department of Faculty Surgery, Dagestan State Medical University, Makhachkala, Russian Federation.
Begova Madina Abdurakhmanovna – student, Dagestan State Medical University, Makhachkala, Russian Federation; madina.begova92@mail.ru, https://orcid.org/0009-0004-3522-7085
Kurbanova Aminat Magomedovna – student, Dagestan State Medical University, Makhachkala, Russian Federation; aminkak27052004@gmail.com, https://orcid.org/0009-0003-7158-8315
Abdurakhmanova Revana Malikovna – student, Dagestan State Medical University, Makhachkala, Russian Federation; revanaabdurahmanova@icloud.com, https://orcid.org/0009-0007-1514-2756
Kurbanova Aysha Khamzatovna – student, Astrakhan State Medical University, Astrakhan, Russian Federation; kurbanova-aysha@list.ru.
Tagirova Fatima Shamilevna – student, Pirogov Russian National Research Medical University, Moscow, Russian Federation; fatima.tagirova@mail.ru, https://orcid.org/0009-0005-4398-8551
Gadzhikhanova Patimat Tatamovna – student, Russian University of Medicine, Moscow, Russian Federation; Gadjikhanova04.06@mail.ru, https://orcid.org/0009-0004-9784-4943
Mutueva Dzhamilya Kamaltinovna – student, Dagestan State Medical University, Makhachkala, Russian Federation; Milya557733@mail.ru, https://orcid.org/0009-0000-8789-1691