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**Dilated Cardiomyopathy in Pediatric Patients with Dystrophic Epidermolysis Bullosa: Retrospective, Cross-Sectional (Registry) Study**

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***Background.*** *One of the most severe forms of epidermolysis bullosa (EB) is its dystrophic form (DEB). This disease is caused by mutations in the COL7A1 gene, leading to skin fragility and blistering, it is characterized typically by severe course of skin lesions and systemic manifestations. One of the challenging systemic complications in patients with DEB is dilated cardiomyopathy (DCM) or secondary myocardial changes aggravating DEB clinical course and significantly affecting morbidity and mortality. However, there is no data on any correlation between DEB severity and anthropometric, osteodensimetry, and laboratory results, as well as with echocardiography (EchoCG) parameters among children with EB in Russian Federation.* ***Objective. The aim of the study is to*** *to analyze the clinical and laboratory profile of patients with DEB and DCM, as well as to identify possible markers associated with high disease severity and risk of mortality in children with DEB in Russian Federation.* ***Methods.*** *The severity of EB course via Birmingham Epidermolysis Bullosa Severity score (BEBS) (from 15.0 to 64.0); anthropometric measures (WAZ, HAZ, BAZ) and bone mineral density status (BMD, Z-score); laboratory parameters (hemoglobin, ferritin, albumin, iron, vitamin D, etc.); EchoCG parameters (ejection fraction (EF), cardiac chambers dimensions, regurgitation); presence of comorbidities and lethal outcomes were evaluated.* ***Results.*** *491 children with EB were registered in Russian Federation according to the data from the "Register of genetic and other rare diseases" of charitable foundation “BELA. Butterfly Children” as at 2025. There is data on 7 patients (4 girls and 3 boys) with severe DEB and comorbid DCM. Patients' age was from 4 to 17 years (mean age 13.4±5.1 years). Higher BEBS values (>50) were associated with severe weight deficit (BAZ<−5), low bone mineral density (Z-score up to −5.3), severe anemia (Hb<90 g/L), and severe myocardial changes (reduced EF, dilated chambers, multiple regurgitations). 3 girls out of these 7 patients died. Tthey had maximum BEBS score (≥46), severe malnutrition, and the most significant changes according to EchoCG.* ***Conclusion.*** *DEB severity (according to BEBS) has negative correlation with anthropometric and osteodensimetry parameters (WAZ, BAZ, Z-score) and is directly related to the risk of severe cardiomyopathy and death. All patients suffering from EB with suspected DCM require complex management, including nutritional support, anemia correction, vitamin D replacement, and regular cardiac monitoring.*

***Keywords****: epidermolysis bullosa, cardiomyopathy, malnutrition, children*

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**Table 1.** Characteristics of patients with DEB and DCM (n=7) as at January 1, 2024

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Indicators** | **Mean** | **Standard deviation** | **Min** | **Max** |
| Age | 12,71 | 5,49 | 4,0 | 17,0 |
| BEBS | 48,21 | 16,23 | 15,0 | 64,0 |
| Hemoglobin | 90,71 | 30,51 | 48,0 | 147,0 |
| Proteinuria | 0,33 | 0,71 | 0,0 | 1,78 |
| Total protein | 68,96 | 34,26 | 0,0 | 92,0 |
| Albumin | 32,99 | 7,04 | 23,0 | 45,29 |
| Cholesterol | 2,59 | 1,44 | 0,0 | 4,08 |
| Creatinine | 30,23 | 18,38 | 15,0 | 69,62 |
| Iron | 5,5 | 6,84 | 1,6 | 20,9 |
| Ferritin | 34,09 | 43,76 | 4,31 | 129,5 |
| Vitamin D | 18,46 | 10,28 | 7,0 | 31,9 |
| *Z*-score osteodensimetry | −2,46 | 2,45 | −5,3 | 1,5 |
| BMD | 0,44 | 0,30 | 0,0 | 0,839 |
| WAZ | −2,85 | 2,14 | −6,58 | −1,23 |
| HAZ | −2,194 | 1,71 | −4,53 | 0,52 |
| BAZ | −3,72 | 1,96 | −6,32 | −1,12 |
| Sodium | 136,15 | 2,98 | 132,0 | 141,0 |
| Magnesium | 0,75 | 0,12 | 0,58 | 0,87 |

**Fig.** Correlation matrix between BEBS and clinical and laboratory parameters

Correlation heatmap

Age / BEBS / Hemoglobin / Proteinuria / Total protein / Albumin / Cholesterol / Creatinine / Iron / Ferritin / Vitamin D / Z-score osteodensimetry / BMD / WAZ / HAZ / BAZ / Sodium / Magnesium

**Table 2.** Left ventricular end diastolic dimension and ejection fraction in children with DEB and DCM

|  |  |  |
| --- | --- | --- |
| **Patient** | **LVEDD, mm** | **LVEF, %** |
| **1** (S.A., 17 years, m) | 57 | 62 |
| **2** (M.Yu., 15 years, m) | 46 | 58 |
| **3** (A.A., 17 years, m) | 52 | 58 |
| **4** (R.D., 17 years, f) | 52 | 58 |
| **5** (D.I., 4 years, f)\* | 43 | 32 |
| **6** (F.M., 6 years, f)\* | 34 | 69 |
| **7** (M.N., 13 years, f)\* | 36 | 67 |
| Min | 34 | 32 |
| Max | 57 | 69 |
| ***Mean* ± *SD*** | 45,7 ± 8,6 | 57,7 ± 12,2 |

*Note.* LVEDD *(*КДР ЛЖ) — left ventricular end diastolic dimension; LVEF (ФВ ЛЖ) — left ventricular ejection fraction. Data from all 7 patients were used to calculate mean and standard deviation (Mean±SD). Patients marked with an asterisk (\*) have lethal outcome.

## RESEARCH LIMITATIONS

This study has number of limitations. The major one is the small sample size of patients with DCM (n=7) which is associated with the rarity of this pathology at EB. All included patients were children under the age of 18 years, that limits the ability to extrapolate the obtained results on adult cohort with DEB and does not allow to fully conclude about the age structure and real mortality at this disease form. Another limitation is the inability to perform full statistical analysis with high degree of confidence other than descriptive and correlational estimation of trends.

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Not declared.

**AUTHORS’ CONTRIBUTION**

**Roman V. Epishev** — study concept, methodology development, data processing, data analysis, conducting th study, visualization, manuscript draft writing, manuscript review and editing.

**Nikolay N. Murashkin** — study management, project administration, study concept, methodology development, conducting th study, manuscript review and editing.

**Olga S. Orlova** — study concept, methodology development, data processing, data analysis, conducting th study, visualization, manuscript draft writing, manuscript review and editing.