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**Fibrous Hands Deformities in Children with Dystrophic Epidermolisys Bullosa (According to the National Register Data)**

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***Background.*** *Dystrophic epidermolysis bullosa (DEB) is one of the most common and severe forms of epidermolysis bullosa (EB). Progressive fibrous tissue remodeling in DEB is characterized by development of limbs contractions and pseudosyndactylies. Complications of DEB have significant negative impact on patients development, adulting, and quality of life. DEB prevalence , its severity, and its correlation with limb deformities has not been examined in Russian studies.* ***Objective. The aim of the study is to*** *examine fibrous hands deformities prevalence and structure in children with DEB in Russian Federation.* ***Methods.*** *The study included data from children with genetically verified DEB (pathogenic variant in the COL7A1 gene) under the age of 18 years with Russian citizenship. Data source is the "Register of genetic and other rare diseases" of Charitable foundation “BELA. Butterfly Children”. Data was collected during the period from September 2014 to June 2025. Fibrous hands deformities were diagnosed in patients with hand pseudosyndactylies and contractions. The severity of single-hand deformities was determined via J. Glicenstein et al. scale (Grade 1–4).* ***Results.*** *Hands deformities of various severity were revealed in 39 (14.9%) out of 261 patients with DEB; 19 (49%) of them were female. The mean age of patients with hands deformities was 12.2±3.6 years (range from 6 to 18 years). Grade 1 deformity (initial deformities) was revealed in 6 (15%) patients, Grade 2 (moderate deformities) — in 23 (59%) patients, Grade 3a (progressive form) — in 9 (23%) patients, Grade 4b (significant changes) — in 1 (3%) patient. The mean age of fibrous hands deformities onset was 4.1±2.9 years. Moreover, its onset occured on average 1.5-2 years earlier in patients with more severe forms (Grade 3a, 4b) than in patients with Grade 1-2.* ***Conclusion.*** *Moderate fibrous limbs deformities (hands) are the most common in the structure of DEB cases. Deformities severity is directly determined by the age of fibrous changes onset.*

***Keywords****: epidermolysis bullosa, dystrophic epidermolisys bullosa, pathological fibrosis, contractions, pseudosyndactylies, TGF-β, fibroblasts, collagen VII, children*

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**Table 1.** Classification of hands deformities at DEB

|  |  |
| --- | --- |
| **Deformity grade** | **Characteristics** |
| Grade 1 (initial deformities) | No nails, proximal fingers phalanges adhesion, no significant grip function violation |
| Grade 2 (moderate deformities) | Thumb adduction, flexion contractions of fingers (shortening of proximal and distal interphalangeal joints ligaments). Fine motor skills impairment, grip function preserved |
| Grade 3а (progressive form) | Adhesion of palmar surface and fingers, thumb adduction. Loss of finger mobility with residual anatomical differentiation. Grip is still possible |
| Grade 3b | Fingers are separated, but gripping is impossible due to adhesions on palmar surface |
| Grade 4а | Complete adhesion, wrist is mobile |
| Grade 4b (significant changes) | Complete adhesion with wrist contraction |

**Table 2.** Patients with DEE and fibrous hands deformities: distribution and age of deformities onset

|  |  |  |  |
| --- | --- | --- | --- |
| **Grade** | ***N*, abs.** | **Age, years\*** | **Age of onset, years\*\*** |
| Grade 1 | 6 | 13,8 ± 1,9 | 4,5 ± 4,3 |
| Grade 2 | 23 | 11,8 ± 3,7 | 4,7 ± 2,8 |
| Grade 3a | 9 | 13,1 ± 4,0 | 2,7 ± 1,1 |
| Grade 4b | 1 | 17,0 | 3,0 |

*Note.* <\*> — mean age; <\*\*> — mean age of fibrous hands deformities onset.

**Fig. 1.** Grade 1: no nails, proximal fingers phalanges adhesion

*Note.* Here and for Fig. 2-4: patients’ parents have signed written informed voluntary consent on the publication of children photos (parts of body) in medical journal (electronic version included) (signed on 19.05.2025).

Source: Epishev R.V. et al., 2024.

**Fig. 2.** Grade 2: thumb adduction, flexion contractions of fingers (shortening of proximal and distal interphalangeal joints ligaments).

Source: Epishev R.V. et al., 2024.

**Fig. 3.** Grade 3a: adhesion of palmar surface and fingers, thumb adduction, grip is still possible

Source: Epishev R.V. et al., 2024.

**Fig. 4.** Grade 3b: fingers are separated, but gripping is impossible due to adhesions on palmar surface

Source: patient's family archive.

**Fig. 5.** Grade 4a: complete adhesion, wrist is mobile

Source: Epishev R.V. et al., 2024.

**Fig. 6.** Grade 4b: complete adhesion with wrist contraction

Source: Epishev R.V. et al., 2024.

**RESEARCH LIMITATIONS**

The research limitation is the retrospective data collection, as it narrows down the possibilities for analyzing the fibrous hands changes progression. Moreover, it should be noted that despite the deformities severity assessment according to the adopted J. Glicenstein et al. scale [8] subjectivity is still possible, and differentiation between 3a and 3b grades as well as transitional forms (such as Grade 4a) may be difficult without comprehensive functional examination.

This study did not include analysis of pathogenic COL7A1 variants, levels of systemic inflammation, and data on rehabilitation or surgical management. It limits data completeness on these factors role in hand deformities development. However, despite the indicated limitations, the obtained results represent significant contribution to the study of fibrous changes structure among children with DEB in Russian Federation, and it can become the basis for further prospective studies with extended clinical and genetic characteristics.

**FINANCING SOURCE**

Not specified.

**DISCLOSURE OF INTEREST**

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.

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