International Journal of Medical Sciences And Clinical Research

VOLUME Vol.06 Issue02 2026
PAGE NO. 163-168
DOI 10.37547/ijmscr/Volume06Issue02-28

Screening and Diagnostic Methods for Undifferentiated Connective Tissue Dysplasia in Children

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Received: 31 December 2025; **Accepted:** 23 January 2026; **Published:** 28 February 2026

Abstract: One of the pressing issues in pediatric pathology is connective tissue dysplasia, particularly its undifferentiated form, which occurs in 70–80% of children. This condition presents significant challenges in the diagnosis and management of comorbid diseases due to the polymorphism of clinical manifestations and variability of phenotypic features. Timely identification and the development of an individualized treatment strategy allow for the prevention of complications and improvement of therapeutic outcomes. Given the high prevalence of undifferentiated connective tissue dysplasia, this pathological condition should be routinely considered during pediatric assessment.

Keywords: Connective tissue dysplasia, diagnosis, screening, phenotypic features, visceral manifestations, cutaneous manifestations.

Introduction: Connective tissue dysplasia is a multifactorial disorder characterized by congenital or familial, heritable defects of connective tissue. It is marked by a combination of external phenotypic features and visceral manifestations, genetic heterogeneity, and a generally favorable clinical course [1].

Researchers of the S.M. Kirov Military Medical Academy conducted comprehensive clinical and screening examinations in more than one hundred pediatric patients. Based on these investigations, fifty phenotypic signs characteristic of the undifferentiated form of connective tissue dysplasia were identified, and a scoring system was developed to assess their severity [2–3]. In constructing this scale, the degree of manifestation of phenotypic features was taken into account, with each parameter scored from 0 to 3 points.

Children admitted to the hospital underwent thorough clinical evaluation; each phenotypic trait was assessed individually, and the cumulative score was calculated. A total score of 30 points or higher was considered diagnostically significant. The scoring scale was developed based on the following criteria: (1) the final cumulative score; (2) the specificity of individual features for the given condition; and (3) the diagnostic coefficient of each feature [4].

Domain of Manifestation	**Clinical Features**	**Scoring Criteria** |	Coefficient

1. Medical History (Anamnesis)	1) Delayed wound and scar healing 2) Joint pain 3) Shortness of breath 4) Rapid fatigue 5) Presence of bruising on the skin 6) Epistaxis (nosebleeds)	0 – no complaints 1 – occurred once in lifetime 2 – recurrent during the year 3 – occurs monthly	1 1 1 1 1 1
2. General Examination	1) Umbilical hernias / abdominal wall folds 2) Muscle diastasis (rectus abdominis separation) 3) Asthenic body habitus 4) Muscle hypoplasia / subcutaneous fat hypoplasia	**1) Umbilical hernias / abdominal wall folds:** 0 – minimal fold 1 – single anterior abdominal wall fold 2 – two or more folds **2) Muscle diastasis:** 0 – absent 1 – mild 2 – moderate 3 – pronounced **3) Asthenic body habitus:** 0 – absent 1 – mild 2 – moderate 3 – pronounced **4) Muscle / fat hypoplasia:** 0 – absent 1 – mild 2 – moderate 3 – pronounced	2 2 3 3
3. Skin Manifestations	1) Atrophic striae / visible venous networks 2) Excessive skin extensibility	**1) Atrophic striae / venous networks:** 0 – absent 1 – mild	1 1

	3) Ecchymoses (bruising) 4) Dry and wrinkled skin 5) Transverse abdominal folds	2 – moderate 3 – pronounced **2) Excessive skin extensibility:** 0 – absent 1 – mild 2 – moderate 3 – pronounced **3) Ecchymoses:** 0 – absent 1 – mild (1–3 petechiae) 2 – moderate (>3 petechiae) 3 – pronounced (multiple petechiae and ecchymoses, up to 3 cm on the abdomen) **4) Dry and wrinkled skin:** 0 – absent 1 – mild 2 – moderate 3 – pronounced **5) Transverse abdominal folds:** 0 – absent 1 – mild 2 – moderate 3 – pronounced	1 1 1
4. Head and Facial Features	1) Dolichocephaly (elongated head) 2) Long or short neck 3) Auricular anomalies (low-set ears, abnormal development, small or large auricles, prominent, folded or misshapen ears)	**1) Dolichocephaly:** 0 – absent 1 – mild 2 – moderate 3 – pronounced **2) Neck length:** 0 – normal 1 – mild deviation 2 – moderate deviation	1 1 1 1

	4) High-arched ("gothic") palate	3 – pronounced deviation **3) Auricular anomalies:** 0 – normal 1 – 1 anomaly 2 – 2–3 anomalies 3 – >3 anomalies **4) High-arched palate:** 0 – absent 1 – mild 2 – moderate 3 – pronounced	
5. Torso / Body Features	1) Chest deformities (pectus excavatum, pectus carinatum) 2) Scoliosis 3) Thoracic kyphosis	**1) Chest deformities:** 0 – absent 1 – mild (grade 1) 2 – moderate (grade 2) 3 – pronounced (grade 3) **2) Scoliosis:** 0 – absent 1 – mild (grade 1) 2 – moderate (grade 2) 3 – severe (grade 3–4) **3) Thoracic kyphosis:** 0 – absent 1 – mild (grade 1) 2 – moderate (grade 2) 3 – severe (grade 3 or higher)	3 3 1
6. Facial Features	1) Hypertelorism / hypotelorism (widely or closely spaced eyes) 2) Ocular pathologies (lens protrusion, keratoconus, anisocoria, blue sclera, colobomas)	**1) Eye spacing:** 0 – absent 1 – mild 2 – moderate 3 – pronounced **2) Ocular pathologies:** 0 – absent	2 2

	3) Mandibular asymmetry	1 – 1 feature 2 – 2–3 features 3 – >3 features **3) Mandibular asymmetry:** 0 – absent 1 – mild 2 – moderate 3 – pronounced	1
7. Upper Limbs / Hands	1) Joint hypermobility 2) Long fingers (arachnodactyly)	**1) Joint hypermobility:** 0 – absent 1 – mild 2 – moderate 3 – pronounced **2) Long fingers:** 0 – absent 1 – mild 2 – moderate 3 – pronounced	3 3
8. Lower Limbs / Legs	1) Flatfoot (pes planus) 2) Knee joint hypermobility	**1)Flatfoot:** 0 – absent 1 – mild (grade 1) 2 – moderate (grade 2) 3 – severe (grade 3 or higher) **2) Knee joint hypermobility:** 0 – absent 1 – mild 2 – moderate 3 – pronounced	2 2

METHODS

The present study was conducted among 179 pediatric patients admitted to the emergency department of 4-SHBKSH. The signs of undifferentiated connective tissue dysplasia were identified and assessed according to the scoring system and diagnostic coefficients presented in Table 1. In this scale, musculoskeletal manifestations were assigned coefficients of 2 and 3, whereas other clinical features were assigned a coefficient of 1.

RESULTS

According to the findings of our study, signs related to

the musculoskeletal system were identified in the vast majority of patients (92.6%). Among these manifestations, joint hypermobility was observed in 68.8% of cases, elongated fingers in 52.7%, scoliosis and kyphosis in 41.9%, flatfoot in 24.7%, and cranial and chest deformities in 17.4% of patients.

Cutaneous manifestations ranked second in frequency. Dry skin, increased skin extensibility, and prominent venous patterns were detected in 72.6% of children, while easy bruising was observed in 52.4% of cases. Other clinical signs accounted for 38.7%.

DISCUSSION

Each clinical feature was scored from 0 to 3 points and subsequently multiplied by the assigned diagnostic coefficient. A total score of 30 points or higher was considered indicative of a high degree of connective tissue dysplasia. The results demonstrate that manifestations of connective tissue dysplasia predominantly involve the musculoskeletal system, followed by cutaneous signs, while other manifestations occur less frequently.

CONCLUSION

The proposed scoring table may be used as a screening tool for the early detection of undifferentiated connective tissue dysplasia. In addition to identifying phenotypic features, this scale facilitates the assessment of the severity of dysplastic changes in affected patients.

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