The name “connective tissue dysplasia” covers a wide range of disorders.

These disorders are caused by a weakness in the connective tissues such as bone, ligaments, tendons and skin. Children with these disorders may have:

 too much joint movement (hypermobility)

 not enough joint movement (joint contractures)

 fragile bones, skin, blood vessels or ligaments

 degenerative joint disease

 short stature

 spinal complications The disorders can be variable because many affected people have a mixture of these different symptoms. Most connective tissue dysplasias follow common patterns of inheritance. These patterns can help genetic counsellors provide families with information about inheritance in their family.

The disorders can be grouped into:

 Ehlers-Danlos Syndromes (EDS)

 Marfan Syndrome and related disorders

 Skeletal Dysplasias

 Brittle Bone Disorders

 MPS Disorders (Mucopolysaccharidoses).

Case series

Thirty-six patients, aged from 18 to 45 years, with lumbosacral radiculopathy associated with connective tissue dysplasia were examined. Detailed neurological examination, X-ray visualization and MRI of lumbosacral spine section, electromyographic assessment were performed. A five-point scale of neuro-vertebrological symptoms, the Numerical Rating Scale (NRS) and the Roland-Morris Low Back Pain and Disability Questionnaire were used.

The results contained own data on the pathogenesis, clinical manifestations and treatment of dorsopathies in connective tissue dysplasias. Inclusion of long-acting pentoxifylline (vasonite) in the combined therapy in patients with dorsopathy associated with connective tissue dysplasia had a positive effect on disease course, decreased pain intensity and improved life activities [1)].

1)
Chukhlovina ML, Chukhlovin AA. [Diagnosis and treatment of dorsopathy in patients with connective tissue dysplasia]. Zh Nevrol Psikhiatr Im S S Korsakova. 2017;117(7):43-46. doi: 10.17116/jnevro20171177143-46. Russian. PubMed PMID: 28805759.