

to 13q12 chromosome. Research indicates that the mutation is at least 1,200 years old and researchers believe that it has first appeared before the migration of Roma from Northwestern India.

The first symptoms of the disease, which affects both sexes, appear between the age of 3 and 12 when patients develop symmetrical girdle, and later on limb dystrophy. Intellectual and coronary functions remain unchanged. The disease takes a similar course to that of the Duchenne Muscular Dystrophy. Approximately 80% become paralyzed between the age of 10 and 15, while in 15% the ability to move is lost between the age of 15 and 20. In 5% of the patients the disease develops more slowly and disability comes between the age of 20 and 30. The average age at which patients become unable to walk is 13 years.

In Bulgaria the disease has been identified in ten regions: Turgovishte, Razgrad, Rousse, Silistra, Dobrich, Veliko Turnovo, Sliven, Yambol, Pernik and Pazardzhik, namely in the following towns and villages Omurtag, Ilyino, Kardam, Goliam Novo, Aprilovo, Lilyak, Draganovetz, and Elenovo; Kubrat, Zavet, Ispcrih, Rakovski, Ostrovo, and Sinya Voda; Senovo; Zarnik; Pchelarovo and Kavarna; Maysko; Sliven, Topolchane, and Gorno Alexandrovo; Yambol and Lozenetz; Pernik and Breznik; Belovo.

5. Congenital Myasthenic Syndrome (CMS), type Ia

Abicht et al. (1999) have conducted a clinical and genetic research of CMS, type Ia, among patients from 11 non-related Roma families from different countries in Southeastern Europe (Hungary, Serbia, Macedonia, Greece, Turkey and Kosovo) and have discovered a genetic mutation in the epsilon subunit of the acetylcholine receptor ($\epsilon 1267\text{dIG}$) in the 17p chromosome.

In Bulgaria the disease has been identified in more than 20 families. The symptoms of the CMS, type Ia, appear immediately after birth or in early childhood and include fluctuating ptosis, poor feeding ability, and respiratory distress, leading sometimes to sudden death. The disease is relatively light and reacts positively to anticholinesterase therapy.

The CMS, type Ia, has been identified in 16 towns and villages in Bulgaria: Rakitovo, Pagnagyurishte, Plovdiv, Lom, Vidin, Pleven, Staroseltzi, Knezha, Kubrat, Dobrich, Sofia, Samokov, Dupnitsa, Maritza, Pirdop, and Elin Pelin. All patients belong to the Yerlii group. The disease has not been discovered in the subgroups of the Wallachian group.

6. Distal Myopathy

Extensive field research identified 21 cases of distal myopathy (12 men and 9 women) in 10 families. The symptoms of the disease appear after the age of 20 when patients start experiencing difficulties walking on their heels, climbing stairs and running. Later on the disease progresses into hand muscle weakness leading to unsteady grip and reduced ability to hold. The onset of the disease varies between the age of 20 and 30, the average age being 23.75 ± 3.59 years. Patients become disabled at an average age of 33.67 ± 3.37 years and lose their ability to walk 9.75 ± 3.55 years after the onset. Electromyographic tests indicate myopathic changes. ECG and ultrasound tests have revealed coronary transformations in 12 out of 21 patients. CAT/MRI tests show that calf and arm muscles are more affected than lumbar muscles.

The disease has been identified in the regions of Vratza, Veliko Turnovo, Dobrich, Varna, and Haskovo, namely in the following towns and villages: Dobrich, Kavarna, and Bozhuretz; Varna and Kamenar; Oriahovo; Tsarksi Izvor; Svilengrad.

7. Spinal Muscular Atrophy – a group of hereditary diseases, which are characterized with degeneration of the peripheral motor neurons leading to progressive paralysis with muscular atrophy. The progress of the disease varies greatly. Spinal muscular atrophy, type 1, leads to severe muscular weakness and hypotonia and strikes infants between birth and six months. Death from respiratory failure usually occurs after the first five years. The first symptoms of spinal muscular atrophy, type 2, appear before the 18th month, while those of type 3 occur