and 25, and bulbar paralysis – between the age of 50 and 60. Gradually thenar and metatarsal muscles atrophy and deformities appear. All patients have a difficulty walking, while older ones are unable to walk. Permanent disability comes after the age of 30. The genetic defect, which causes the disease (Kalaydjieva et al., 2000), has been identified and genetic prophylaxis may be introduced.

The disease has been identified in 40 towns and villages: in the endogamous Wallachian subgroup in Mladenovo and Humata Districts in Lom and in Montana; among the Kardarashi in Rousse, Cherven Briag, Pernik, Levski, Kostinbrod, Bukovluk, Gorna Mitropolia, and Rudartzi; among the Kopanari in Dushevo, Yavoretz, Shumata, Sadovetz, Gradina, Gorni Dubnik, Pravetz, Koynare, Sofia, Tvurditza, Shivachevo, and Kamenovo; among the Dzambazi in Nedan and Ovcha Mogila; among the Burgudzi in Kamen and Viatovo; among the Grebenari in Ivaylo and Glavinitza; among the Bulgarian-speaking Yerlii in Panagyurishte and Stara Zagora; among the Turkish-speaking Yerlii in Pazardzhik, Samokov, Etropole, Velingrad, Plovdiv, Dobrich, Silistra, Balchik, and Pleven.

Later on the disease has also been identified among Roma in other European countries: Italy (Merlini et al., 1998), Slovenia (Butinar et al., 1999), Germany (Baethmann et al., 1998), Spain (Colomer et al., 2000), France, Romania, Belgium (Kalaydjieva et al., 2000).

2. Congenital Cataract Facial Dysmorphism Neuropathy Syndrome – a dysontogenetic neurodegenerative disease affecting both central and peripheral nervous system, which has been mapped to 18q23 chromosome (Angelicheva et al., 1999). The disease is first described by Turnev et al. in 1999. The main symptoms among all patients are congenital zonular cataracts and microcornea, facial dysmorphism and nanism, neuropathy with peripheral paralysis, mental disabilities with cerebral atrophy and hydrocephaly, reduced bone mass, skeletal deformations and disturbed sexual development.

The disease has been identified among the Kopanari in the villages Liliache, Hurletz, Boychinovtzi, Sadovetz, Zdravetz, Podgoritza, Rossena, Markovo, Brestoviane, Tvurditza, Kamen, Shumata, Yulievo, Stozher, Smirnenski, Novo Selo, and Sindel, as well as in the Kremikovtzi and Botunetz Districts in Sofia.

The disease has been identified among other Roma groups and subgroups: the Wallachian Dzambazi in Portitovtzi, Kozlovetz, Strahilovo, and Ovcha Mogila; among the Kardarashi in Rousse; among the Burgudzi in Kamen; among the Bulgarian-speaking Yerlii in Karlovo, Sopot, and Shoumen; among the Turkish-speaking Yerlii in Kamenar, Aytos, Exarch Antimovo, Krustina, Kosharitza, Zabernovo, Roussokastro, Rudnik, and Gulabovo.

The disease has been also identified among Roma in Romania, Hungary, Italy, Germany, and the United States.

3. Hereditary Motor and Sensory Neuropathy, Rousse type is an axonal neuropathy. All patients belong to the strictly endogamous group of the Kardarashi. Most cases have been identified in Rousse, hence the name of the disease. The first symptoms appear between the age of 8 and 14 when patients develop distal paralysis and severe sensory loss. Permanent disability comes around the age of 40. The genetic defect, which causes the disease, has been mapped to 10q23 chromosome (Rogers et al., 2000).

The Hereditary Motor and Sensory Neuropathy, Rousse type, has been identified in only 7 villages and towns: Rousse, Prossena, Pazardzhik, Cherven Briag, Hurletz, and Aksakovo. Later on the disease has also been discovered among Roma in Romania and in Spain.

4. Muscular Dystrophy Gamma-sarcoglycanopathia

Piccolo et al. (1996) have researched a severe autosomal recessive muscular dystrophy with gamma-sarcoglycan deficiency in seven large non-related Roma families in different European countries (France, Spain, Italy and Portugal). They have discovered that the genetic defect maps