

after the 18th month. Inheritance is autosomal recessive. The genetic defect is mapped to the 5 chromosome.

Spinal muscular atrophy, type 1, was identified among Turkish-speaking Roma in Karnobat Bourgas, Assenovgrad, Pazardzhik, Zavet, Isperih, Shoumen, Sliven, and Varna, as well as in the villages Enitza, Samuilovo, and Ostren. Spinal muscular atrophy, type 2, was identified in the villages Ognianovo and Satovcha, while type 3 was discovered in Shoumen, Novi Pazar, Lukovit, Biala Slatina, Koynare, Sliven, Stara Zagora, Pavel Bania.

8. Hepatolenticular Degeneration (Wilson's disease) is an autosomal recessive disorder of copper metabolism characterized by excessive accumulations of copper in the liver, central nervous system, kidneys, and cornea, as well as by liver deficiency, tremor, speech disorders, muscular rigidity, seizures, and dementia. The disease was identified among Roma in the regions of Shoumen, Turgovishte and Razgrad.

9. Primary epilepsy is common among two Roma groups: the Kalajdzii and the Kardarashi. The Kalajdzii are an endogamous group, which lives in the regions of Plovdiv, Stara Zagora, Haskovo, Yambol, Sliven and Bourgas. Research showed that 30% of the Kalajdzii suffer from epilepsy. The disease is also common among Kardarashi in Slunchevo, Ignatievo, Aksakovo, Silistra, Peturch, and Yana.

10. Other hereditary diseases identified during the research include: neurofibromatosis, Strumpel disease, hereditary ataxia, pigmentary retina degeneration, congenital glaucoma, galactosemia, leukodystrophy, hereditary angioedema, mucoviscidosis, congenital arthrogryposis, and others.

A broad epidemiologic research conducted across Bulgaria revealed that hereditary neurologic diseases are quite common among Roma – 55 cases per 100,000 people. Most often such diseases occur in the Wallachian group – the Kalajdzii, the Kardarashi, the Kopanari, the Dzam-bazi and others.

Prophylaxis of Hereditary Diseases

The diagnosing of hereditary diseases among Roma and the discovery of the genetic defects, which cause them, created conditions for conducting prophylactic programs targeted to the risk groups.

The goal of these programs is to help reduce the incidence of hereditary diseases by organizing individual prophylaxis of healthy young adults from the risk groups, identifying carriers, providing genetic consultations, conducting prenatal diagnostics of families in which both partners are carriers.

A pilot genetic program for the prevention of severe muscular dystrophy gamma-sarcoglycanopathia, which has been approved by the Ministry of Healthcare and was financed by the French Anti-Myopathy Association, was introduced in Omurtag. The program was carried out by the Minority Health Problems Foundation – Sofia in cooperation with Associate Prof. Lyuba Kalaydjieva from the Edith Cowan University in Western Australia. The research revealed that 6% of the people in reproductive age carry the disease. A similar program supported by the Open Society Foundation – Sofia was realized in 1999–2000 in Sliven in cooperation with the Roma Health Foundation – Sliven. The research conducted in the Roma community of Nadezhda District in Sliven revealed that 8.4% of the people have this genetic defect.

In 2000 the Council of Ministers with its decision № 535 of November 10, 2000 approved a National Program for Genetic Prophylaxis of Hereditary Diseases and Congenital Anomalies for 2000–2005, which also targets genetic diseases typical for the Roma community. In 2001 the European Neuro Muscular Center organized an international workshop in Naarden, the Netherlands, on Neuromuscular Disorders in the Roma Population, which was attended by prominent European experts. The participants at the workshop discussed diagnostic criteria and ways