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Historical paper

The Meryon Lecture at the 18th Annual Meeting of the Meryon Society Wolfson College, Oxford, UK, 12th September 2014 Neuromuscular disorders in Roma (Gypsies) – collaborative studies, epidemiology, community-based carrier testing program and social activities

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1. Historical background

The Roma (Gypsies) are a transnational minority with an overall population size estimated between 10 and 15 million [1]. There are 8 million Roma in Europe, with close to 70% resident in Central and Eastern Europe, mainly in the Balkans. Data provided by the social sciences and genetic research suggest that Roma represent a conglomerate of genetically isolated founder populations of common origin who have subsequently split into multiple social divergent and geographically dispersed Gypsy groups [2]. Ethnolinguistically the Roma share a common Hindi/Sanskrit core language [3,4]. However, the more than 100 dialects of Romani illustrate the divergent paths that the various Roma groups have taken in their travels [4]. Traces of Middle Eastern languages, Armenian, Persian, Turkish and Kurdish are found in the various dialects [3]. Roma are heterogeneous in nature, changing with and adapting in new cultures and places as they travel [5–7]. The arrival of the Gypsies in the Byzantine Empire is estimated to have occurred, in the 11th–12th centuries [2,8,9], at which point a large fraction of the population settled permanently in the Balkans. The formation of the present-day Romani populations of European countries is the compound product of the early migrations from the Balkans into Western Europe, completed by the 15th century, and three superimposed migration waves: the first during the end of the 19th century, after the abolition of Gypsy slavery in Romania [8,10]; the second out of Yugoslavia, during 1960s and 1970s; and the third during the last decade following the political and economic changes in Eastern Europe [2,11].

The earliest written record of the Roma comes from European town logs of the fourteenth century [3,12]. Historical writings are similar in many ways to the accounts of Roma travels today and describe the varied cultural conflicts that developed between settled peoples and nomadic cultural groups. The dominant European cultures of the early fourteenth century were devoutly Roman Catholic. The fortune telling, begging and nomadic ways of the Roma were looked down upon by settled Christians [3,4]. The Roma of Europe had consistently encountered punitive laws designed to persecute “wanderers” [5]. The history of repression and ethnic discrimination was worst in Romania where Roma peoples spent more than 600 years in slavery from the fourteenth to the mid-nineteenth century. Depending on the owner, Gypsy slaves were split into categories with different degrees of freedom and “privileges”. Within each category, further subdivisions were based on trade, with some Romani groups retaining their professional ethnonyms (e.g. basket-weaves, spoon-makers) to the present day [4,10,12–14]. The persecution of Roma people culminated in twentieth century when most of this population endured World War II in concentration camps such as Auschwitz and Buchenwald. Roma people were concentrated into special sections within these camps for both medical experimentation and extermination [4,5,10,12–15]. This mistreatment left an

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understandable legacy of alienation and mistrust between Roma groups and settled Europeans. At present time in many European countries the policy of assimilation, exclusion and stigmatization continues. Roma meet discrimination and racism which lead them into poverty and to a life on the margin. This reinforced in the Roma the deep cultural distrust of outsiders, alienating them from any desire to interact or borrow anything associated with gajo ways [12].

2. Social organization

The traditional social organization of the Gypsies, still strongly preserved in the Balkans, is based on multiple endogamous groups, similar to the professional *jatis* of India [8,9,11,16]. Liegeois [1] describes the current social organization of the Roma as a "fluid mosaic of diversified groups". Group identity of the ensuing divisions and rules of endogamy are based on a variety of criteria such as tradition, customs and organs of self-rule, language and dialects, trades, history of migrations and religion. Individual groups can be classified into several major metagroups: the Roma of East European extraction including Vlax, Xoroxane (Muslim) and others; the Sinti, long resident in German-speaking lands and known as the Manouches in France and Catalonia; the Kale in Spain, Cigano in Portugal and Gitans of Southern France; and the Romanichals of Britain [11].

The greatest diversity exists in The Balkans, e.g. in Bulgaria, where the 800,000 Roma form three metagroups: Jerlii (with three major divisions: Dassikane, Xoroxane and Vlahichki), Kalderas and Rudari. Each comprises numerous small groups with different rules of endogamy [9,11].

3. Present status of Roma in an expanding Europe

Roma are the most prominent poverty risk group in many of the countries of Central and Eastern Europe. There is a housing segregation as a form of geographic exclusion. In the most of Roma homes there is a lack of basic sanitary conditions. Roma neighborhoods are frequently extremely overcrowded and destitute. Some Roma slums have evocative nicknames; for example, "Abyssinia" and "Cambodja" are extremely impoverished areas within Roma ghettos. Lack of water, gas, electricity, and public services such as waste collection bedevils many Roma neighborhoods. Unemployment rates in Bulgarian Roma vary between 50% and 80%. Because of their low education levels, Roma were most frequently employed in low-skilled manufacturing industries. Roma children often are excluded from education in mainstream public schools in Central and Eastern Europe and instead relegated to schools for the mentally retarded. The different surveys demonstrate lower educational attainment among Roma. Most Roma have primary education or below. In Bulgaria 89% of Roma had primary education and only 10% had some secondary education. Only 1% of Roma continued past secondary school. Healthcare reform in Bulgaria brought to light some very alarming tendencies in Roma health: high morbidity, high mortality, low life expectancy. The average life expectancy among Roma is more than 10 years lower than the average for the country. Percentage of Roma with some kind of disability is 6 times higher than in the Bulgarian population. The lack of basic sanitary conditions is one of the main reasons for the higher incidence of infectious diseases among Roma, compared to the rest of the population. 50% of the Roma families have a chronically ill family member, and 20% have two [17,18]. The tradition of large families and early marriages places women and young girls at increased health risk. The lack of adequate information and knowledge among Roma is one of the main reasons for the discriminatory attitude of unscrupulous general practitioners and hospital staff. The cultural differences and traditions, which are observed piously in some Roma communities, also affect the health condition of Roma. Endogamy and inbreeding, which are most common among subgroups of the Wallachian group, lead to the accumulation of hereditary disorders.

4. Mendelian neuromuscular disorders of the Gypsy caused by private founder mutations

Similar to other genetically isolated founder populations, such as the Finns and the Ashkenazi Jews, the Roma harbor a number of unique or rare autosomal recessive disorders caused by "private" founder mutations. Oppressive policies of persecution, exclusion, containment and forced assimilation practiced toward the Roma in most, if not all, European countries, together with Roma's adherence to an ancient social tradition, have acted together to result in endogamy and isolation, making the Roma one of Europe's largest genetic isolates [11].

As a result of the traditionally low socio-economic status and limited access of the Roma to health care, their unique genetic heritage has long escaped the attention of European medicine. In 1996, after publishing of two papers [19,20], on Limb girdle muscular dystrophy type C due to a private C283Y Gypsy mutation and on a novel disorder – Hereditary motor and sensory neuropathy type Lom, Prof Jean-Claude Kaplan initiated with the support of AFM research activities in the different countries and stimulated European collaboration in studies.

International collaboration has made a substantial contribution to the study of these diseases. In 2001 and 2004, International Workshops of the European Neuromuscular Centre entitled "Neuromuscular Disorders in Roma" were held. During the 125th ENMC Workshop, specialists from Europe, Australia and Japan discussed the diagnostics and prevention of these disorders and future collaboration in improving health care [21].

Collaborative studies have identified novel single-gene disorders and private mutations among the Roma drawing attention to these previously ignored founder mutations: LGMD2C due to mutation C283Y in *SGCG*; CMS due to mutation 1267delG in *CHRNE*; HMSN Lom due to R148X in *NDRG1* gene; CCFDN due to IVS6+389C>T mutation in *CTDP*; HMSN Russe due to a mutation in an alternative untranslated exon of *hexokinase 1* gene; GNE myopathy due to p.Ile587Thr mutation in the *GNE* gene (Table 1). The identification of a number of novel Mendelian disorders and private mutations in the Roma (Gypsies) points to their unique genetic heritage [2].