Epidemiology of Rare Diseases in Cyprus

Background: Huntington disease (HD) and Amyotrophic lateral sclerosis (ALS) are rare progressive neurodegenerative diseases. The epidemiology of ALS and HD in Cyprus, an island in Southern Europe with extensive western European colonization during the past millennium, has never been examined and this was the aim of this study.

Methods: All registered Cypriot ALS patients in the Republic of Cyprus from January 1985 until December 2014 and all registered HD patients in the Republic of Cyprus, since 1994, were included. Sociodemographic and clinical information were recorded and maps, showing the geographic distribution of the disorders, were constructed.

Results: The study identified 179 ALS patients, seven of whom had a positive family history. The mean age at onset was 58.6 years and a slight male predominance was observed. Average annual crude incidence was 1.26 cases/100,000 person-years and at the beginning of 2015, prevalence of ALS was 7.9 cases/100,000 population. Both incidence and prevalence displayed an increasing trend, even after age-standardization of incidence rates. For HD, the project identified 58 clinically manifested cases of HD belonging to 19 families. Sixteen families were of Cypriot origin and were concentrated in a confined geographical cluster in Southeast Cyprus. In 2015 prevalence of symptomatic HD was 4.64/100,000 population, while incidence was 0.12/100.000 person years.

Conclusions: Incidence, prevalence and main sociodemographic characteristics of ALS and HD in Cyprus were similar to those of other European countries. For ALS, an increased incidence through the years was confirmed. For HD, the geographical clustering of HD families observed supports the possibility for a relatively recent founder effect of HD in Cyprus, which could potentially be of western European origin.