

VIII GIORNATA DELLO SPECIALIZZANDO IN NEUROLOGIA

Catania, 11 Giugno 2019

THE PREVALENCE OF HUNTINGTON'S DISEASE IN SARDINIA

Margherita Sechi

*Department of Medical Sciences and Public Health
University of Cagliari, Sardinia, Italy*



PREVALENCE OF HUNTINGTON'S DISEASE IN SARDINIA, ITALY

Accurate estimates of prevalence

- are important for health services planning, given the significant burden of HD on affected individuals
- may become increasingly important in the near future when the results of clinical trials for drug therapy in HD will become available.

**MEAN PREVALENCE OF HUNTINGTON'S DISEASE
IN CAUCASIANS**

5.6 cases / 100.000

Region, year of prevalence	Population size	Prevalence per 100.000 population
<i>Europe</i>		
Finland, 2010	5.337.358	2.14
Iceland, 2007	311.114	0.96
Northern Ireland, 2001	1.698.113	10.6
UK, 2010	4.683.669	12.3
The Netherlands, 2000	15.930.000	6.50
Italy, 2013 (Modena – Reggio)	1.250.000	2.48
Italy, 2013 (Molise)	313.341	10.85
Greece, 2008	10.964.020	5.42
Spain, 2014	660.000	5.00
Slovenia, 2006	2.011.614	5.17
<i>North America</i>		
Canada, 2012	4.609.659	13.7
<i>Asia</i>		
South Korea, 2013	51.141.463	0.41
Taiwan, 2007	23.000.00	0.42
Japan, 2015	1.387.000	0.70

MEAN CAG LENGHT ON NORMAL CHROMOSOMES

Region	Men CAG length \pm SD
Europe	18.4 \pm 3.5
China	16.4 \pm 1.5
Japan	16.6 \pm 1.5
Africa	16.2 \pm 2.5

GEOGRAPHICAL DISTRIBUTION OF HAPLOTYPES AT RISK

Haplotype	Geographic features
High risk A1, A2	> Europe, absent in East Asia
Low risk B, C	> East Asia

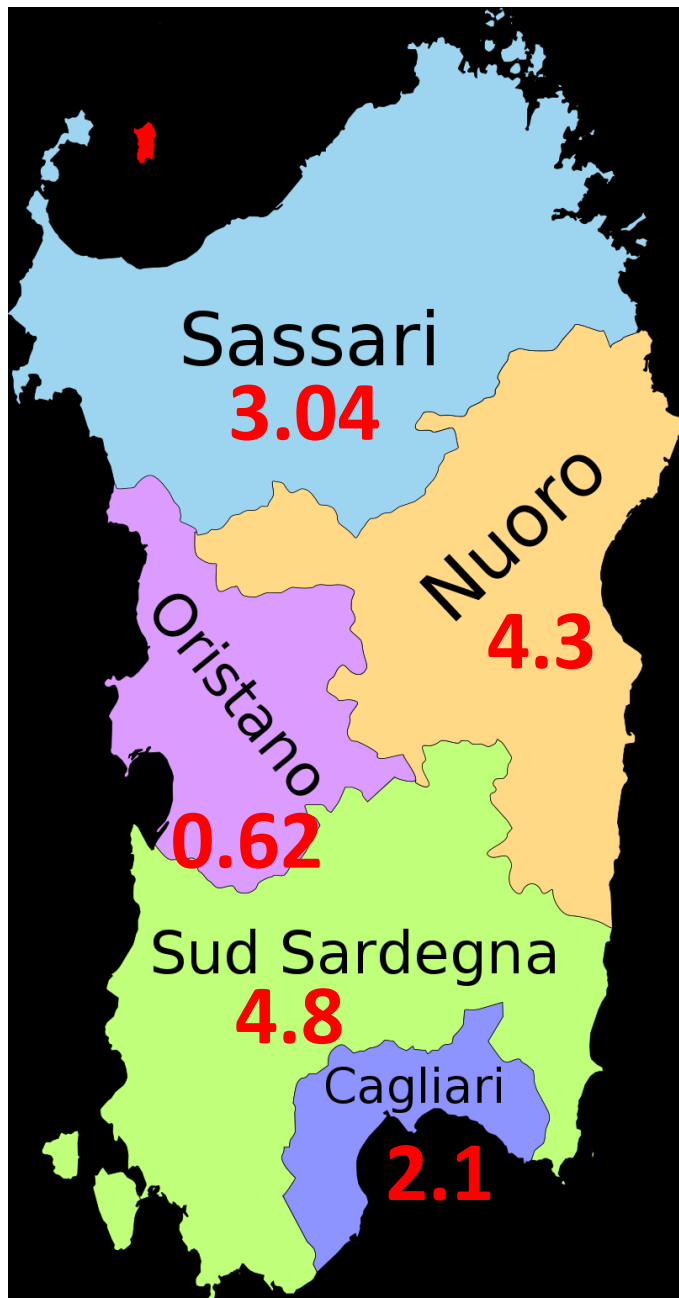
Region, year of prevalence	Population size	Prevalence per 100.000 population
<i>Europe</i>		
Finland, 2010	5.337.358	2.14
Iceland, 2007	311.114	0.96
Northern Ireland, 2001	1.698.113	10.6
UK, 2010	4.683.669	12.3
The Netherlands, 2000	15.930.000	6.50
Italy, 2013 (Modena – Reggio)	1.250.000	2.48
Italy, 2013 (Molise)	313.341	10.85
Greece, 2008	10.964.020	5.42
Spain, 2014	660.000	5.00
Slovenia, 2006	2.011.614	5.17
<i>North America</i>		
Canada, 2012	4.609.659	13.7
<i>Asia</i>		
South Korea, 2013	51.141.463	0.41
Taiwan, 2007	23.000.00	0.42
Japan, 2015	1.387.000	0.70

SOURCES OF PREVALENT CASES

Reference point
for genetic testing
(Binaghi Hospital)

Local
Neurological
services (n.10)

Major neurological
services in Italy
(n. 6)



PREVALENCE OF HUNTINGTON'S DISEASE IN SARDINIA, ITALY– PRELIMINARY RESULTS

	Population (31 /12/ 2017)	Patients (n.)	Prevalence x 10 ⁵ population
Sardinia	1648176	51	3.1
Cagliari	431955	9	2.1
South Sardinia	353830	17	4.8
Oristano	159218	1	0.62
Nuoro	210531	9	4.3
Sassari	492642	15	3.04

MEAN CAG LENGHT ON NORMAL CHROMOSOMES

Region	Mean CAG length \pm SD
Europe	18.4 \pm 3.5
China	16.4 \pm 1.5
Japan	16.6 \pm 1.5
Africa	16.2 \pm 2.5
Sardinia	18 \pm 2.6

MAY PREVALENCE DATA FROM SARDINIA BE UNDERSTIMATED?

Several categories of patients can not have been correctly diagnosed and captured in the prevalence study:

- Patients presenting with atypical manifestations of HD
- Very elderly patients with senile chorea (only one patient in our database was 80s)
- Patients with no family history, and those with new mutations.
- Less common cases in which psychiatric symptoms preceded the movement disorder.

CONCLUSIONS

- Estimates from this preliminary study should be considered as “minimum prevalence estimates”
- Crude prevalence rates were remarkably higher in the inner part of Sardinia than in the other counties, thus suggesting a *founder effect*.

GRAZIE PER L'ATTENZIONE!