MOLEcular AND EPIDEMIOLOGICAL CHARACTERIZATION OF A GEOGRAPHICAL CLUSTER OF HUNTINGTON’S DISEASE IN A BRAZILIAN TOWN OF ZONA DA MATA, MINAS GERAIS STATE, BRAZIL

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Huntington's disease (HD) is an autosomal dominant, neurodegenerative disease, caused by expansions of CAG repeats in the HTT gene, located on 4p16.3. Our aim was to investigate a geographical cluster of Huntington’s disease in Ervalia, a Brazilian town of Minas Gerais State (MG). A descriptive cross-sectional study was performed, starting in January 2011 until June 2013. Therefore, we calculated the minimum prevalence of Huntington’s disease (HD) in Ervalia, known to have many HD affected families. We also determined the genetic profile of the polymorphic CAG region of the HTT gene in 32 subjects of four affected families. PCR and capillary electrophoresis were used for sizing the repeats. The molecular survey (n = 32) showed that the mean number of CAG repeats was 26.89 (±12.71), and the mean current age of the individuals, as of June 1, 2013, was 44.67 years (±17.54). The mean age for the onset of the symptoms (n=20) was 42.67 years (±12.78). The statistical analysis performed between the variables of age of disease onset and the number of expanded alleles showed a strong negative correlation [Pearson (r)= -0.89], with p <0.001. The number of CAG repeats ranged from 14 to 54 for the 32 subjects studied including chromosomes with normal alleles and those with expanded alleles. Regarding the expanded alleles for HD (n= 20), the number of CAG repeats ranged from 40 to 54, with a mean value of 43.52 ± 3.84 and median of 44. In 50% of the cases, individuals carrying expanded alleles had 44 or more CAG copies. Among the expanded alleles, 20 (100%) had a full penetrance (>39 copies). The minimum prevalence of HD in Ervalia was at least 10.3 - 14.4 fold greater than that of the world population, although it does not represent the overall prevalence of the disease in Brazil.

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