CTG repeats at the myotonic protein kinase gene in a healthy Chilean population sample

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To study the variability at the myotonic dystrophy protein kinase (DMPK) gene in a Chilean sample of healthy people. DM1 is an autosomal dominant disorder caused by an expansion of a (CTG) repeat at the 3?-UTR of the gene DMPK. Healthy individuals have alleles under 35 repeats and diseased individuals have over 50. Methods - Genotyping the number of (CTG) repeats at this gene in a sample of healthy Chilean people. Results - Allele frequencies were significantly different from those of other populations. The most frequent allele was with five repeats. The frequency of larger alleles (>18 CTG repeats) was 11%, close to the European frequency (12%) and higher than the Japanese (8%) and Aboriginal Pehuenche samples (8%). Conclusions - Allelic frequencies in the Chilean sample studied were intermediate between those of the two ancestral populations (European and Pehuenche).