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Abstract.

Mutations in *CHCHD10* have been reported as the cause of a large panel of neurological disorders. In order to confirm the contribution of this gene to amyotrophic lateral sclerosis (ALS) disease we analyzed the 4 coding exons of *CHCHD10* by Sanger sequencing in a cohort of 118 French familial ALS already excluded for all known ALS related genes. We did not find any pathogenic mutation suggesting that *CHCHD10* is not a major genetic cause of familial ALS, in France.

Introduction.

CHCHD10 gene encodes a coiled-coil helix coiled-coil helix protein involved in the maintenance of mitochondrial organization, which is crucial for the function of neurons. Indeed, mutations in *CHCHD10* have been identified in broad neurological diseases affecting peripheral or central nervous system neurons including axonal Charcot-Marie-Tooth Type 2 (Pasanen, et al., 2015), late-onset spinal motor neuronopathy (Penttila, et al., 2015), early-onset dementia (Dobson-Stone, et al., 2015), Parkinson's and Alzheimer's diseases (Zhang, et al., 2015).

A *CHCHD10* mutation (c.176C>T, p.Ser59Leu, S59L) was recently identified by whole-exome sequencing analysis in a French family with a mitochondrial DNA instability disorder (Bannwarth, et al., 2014). The eight affected patients carrying this mutation presented a large panel of clinical phenotypes including cerebellar ataxia, motor neuron disease, myopathy, bulbar syndrome, sensorineural deafness and cognitive decline resembling dementia (Bannwarth, et al., 2014). The S59L mutation was further identified in 1 out of 21 families with pathologically proven frontotemporal dementia/amyotrophic lateral sclerosis (FTD/ALS) (Bannwarth, et al., 2014). FTD is characterized by a degeneration of frontal and temporal lobe neurons leading to personality and behavior changes and/or language impairment. ALS is characterized by degeneration of central and peripheral motor neurons leading to a rapidly fatal paralysis. FTD is diagnosed in 13-14% of ALS patients (Montuschi, et al., 2015; Phukan, et al., 2012) and 12-14% of patients with FTD secondarily develop ALS (Burrell, et al., 2011; Lomen-Hoerth, et al., 2002).

Several studies have documented the identification of *CHCHD10* mutations in FTD/ALS (Chaussenot, et al., 2014; Dobson-Stone, et al., 2015; Dols-Icardo, et al., 2015), FTD (Dols-Icardo, et al., 2015; Zhang, et al., 2015), or pure ALS patients (Abdelkarim, et al., 2015; Chio, et al., 2015; Dols-Icardo, et al., 2015; Johnson, et al., 2014; Kurzwelly, et al., 2015; Muller, et al., 2014; Ronchi, et al., 2015; Zhang, et al., 2015). In contrast, other studies did not find any pathogenic variant in ALS patients despite frequent mutations being identified in pure FTD patients from the same geographic area (Jiao, et al., 2015;

Marroquin, et al., 2015; Wong, et al., 2015). In France *CHCHD10* is responsible for 2.6% of FTD/ALS cases (Chaussenot, et al., 2014). We aimed to determine the *CHCHD10* mutation frequency in a large cohort of French familial ALS (FALS) patients.

Methods.

A cohort of 118 FALS was analyzed in this study. FTD was secondarily diagnosed in 15% of these ALS patients. Patients were previously analyzed for main ALS related genes (supplementary material). The 4 *CHCHD10* coding exons (ENST00000484558) were analyzed by Sanger sequencing. Sequence of the primers and PCR conditions are available upon request.

Results.

We identified four variants in *CHCHD10* in our population of ALS patients: three synonymous at the heterozygous state (c.141G>A, p.Gln47Gln, Q47Q; c.234G>A, p.Ser78Ser, S78S; c.330G>A, p.Leu110Leu, L110L) and one missense at the homozygous state (c.286C>A, p.Pro96Thr, P96T). This substitution is predicted to activate a cryptic acceptor site which could impact on *CHCHD10* splicing (Netgene2, http://www.cbs.dtu.dk/services/NetGene2/). In silico analyses predicted that the P96T amino acid change was possibly damaging (PolyPhen-2, http://genetics.bwh.harvard.edu/pph2/) or deleterious (SIFT, http://sift.bii.a-star.edu.sg/) for the protein functions. Three of the variants we identified (S78S, P96T and L110L) were already reported in public databases (Supplementary Table 1). The L110L variant, which is more frequent in African than in European population (Supplementary Table 1), was identified in a patient from Martinique origin (Supplementary Table 2). The patient carrying the P96T variant at the homozygous state had a slow ALS disease duration of 108 months (Supplementary Table 2).

Discussion.

The analysis of 118 French ALS patients identified three synonymous variants that are not likely to be pathogenic. We also identified the missense P96T variant at the homozygous state in a patient presenting a slow progression of the disease (9 years). Similar slow disease duration (6 to 17 years)

3

was previously observed for patients carrying R15L, G66V or P80L *CHCHD10* variants (Muller, et al., 2014; Zhang, et al., 2015). This P96T variant was predicted to impact *CHCHD10* splicing and to have a deleterious effect on protein function, supporting the view that it could be pathogenic. Interestingly, it was already identified at the homozygous state in 2 Spanish ALS patients (Dols-Icardo, et al., 2015). However this variant was also previously identified in Italian ALS patients and controls (Chio, et al., 2015) and is more frequent in some specific populations: frequencies are 0.02533 (ExAC) and 0.005 (1000 Genomes project) for Latino and Iberian Spanish controls, respectively. Thus, as for the P34S variant, which was identified at a similar frequency in patients and controls, (Abdelkarim, et al., 2015; Dols-Icardo, et al., 2015; Marroquin, et al., 2015; Wong, et al., 2015), the pathogenicity of the P96T variant should be interpreted with caution and is likely not sufficient to cause ALS. Overall, as none of our variants seemed to be pathogenic, we concluded that *CHCHD10* is not a primary cause of FALS in France.

Disclosure statement.

The authors declare no actual or potential conflicts of interest.

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Supplementary material.

Methods.

The diagnosis of ALS was based on the El Escorial criteria established by the World Federation of Neurology (Brooks, et al., 2000) and that of FTLD was based on international diagnosis criteria (Rascovsky, et al., 2011). Criterion for family inclusion was that at least two members were affected by ALS. FALS patients were devoid of mutation in *C9ORF72, SOD1, TARDBP, FUS, VAPB, ANG, UBQLN2, VCP* or *PFN1* (Millecamps, et al., 2012a; Millecamps, et al., 2012b; Millecamps, et al., 2010b). These FALS patients had also systematically been screened for *DAO, OPTN, SQSTM1, SS18L1* and *MATR3* (Millecamps, et al., 2011; Millecamps, et al., 2010a; Millecamps, et al., 2014; Teyssou, et al., 2013; Teyssou, et al., 2014) mutations. However patients carrying the variants we previously reported in these later genes were included in this study as the pathogenicity of these variants still remains unclear. Mean age of onset of the analyzed series was 60 years (SEM 1, range 33 to 80 years) and mean disease duration was 48 months (SEM 6, range 6 to 260 months). ALS patients were evaluated with standardized behavioral procedures for cognitive impairments whenever this was possible, considering the rapid progression of the disease in these patients.

Variant	rs number	ExAC	ESP	1000 Genomes
c.286C>A, p.Pro96Thr	rs111677724	Eur: 0.000998 Afr: 0. 2559	EA: 0 AA: 0.015	Eur: 0 Afr: 0.057
c.330G>A, p.Leu110Leu	rs138183274	Eur: 0 Afr: 0.0085	EA: 0 AA: 0.0073	Eur: 0 Afr: 0.012
c.234G>A, p.Ser78Ser	rs111527940	Eur: 0.01314 Afr: 0.003376	EA: 0.0101 AA: 0,0025	Eur: 0.010 Afr: 0
c.141G>A, p.Gln47Gln	/	/	/	/

Supplementary Table 1. Frequency of heterozygous *CHCHD10* variants in control populations.

ExAC: Exome Aggregation Consortium (http://exac.broadinstitute.org), ESP: the NHLBI Exome Sequencing Project (http://evs.gs.washington.edu/EVS), 1000 Genomes project (http://www.1000genomes.org/), Eur: European (non-Finnish), Afr: African, EA: European American, AA: African American.

Variant	Sexe	Origin	Age of onset	Site of onset	Disease duration	Remark
P96T	F	French	53 y	upper limb (distal)	108 months	Predominant lower motor neuron involvement
L110L	F	Martinique	43 y	lower limb (distal)	na	
S78S	M	French	68 y	upper limb (distal)	19 months	ALS-FTD
Q47Q	F	French	59 y	lower limb (distal)	15 months	

Supplementary Table 2. Clinical data of ALS patients carrying a *CHCHD10* gene variant.

F: Female / M: Male / y: years / na: not available.

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