confirmou o diagnóstico de MPS VI, um distúrbio do armazenamento lisossomal raro com um padrão de herança autossómica recessiva.

**Conclusão**: Os nossos resultados sublinham a importância do reconhecimento do ESP como uma ferramenta diagnóstica valiosa, que fornece informação relevante para o diagnóstico de diversos distúrbios hereditários hematológicos e não-hematológicos.

**P-19**

## FRONTOTEMPORAL DEMENTIA AND NEURONAL CEROID LIPOFUSCINOSIS

*Ana Luísa Carvalho[1], Lina Ramos[1], Maria Margarida Venâncio[2], Isabel Santana[3], Carmo Macário[4], Rosário Almeida[5], Jorge Saraiva[6]*

*[1] Medical Genetics Unit, Hospital Pediátrico, Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal*

*[2] Medical Genetics Unit, Hospital Pediátrico, Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal and Department of Medical Genetics, Faculty of Medicine, University of Coimbra, Portugal*

*[3] Department of Neurology, Hospitais da Universidade de Coimbra, Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal and Faculty of Medicine, University of Coimbra, Portugal*

*[4] Department of Neurology, Hospitais da Universidade de Coimbra, Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal*

*[5] CNC, Center for Neuroscience and Cell Biology, University of Coimbra, Portugal*

*[6] Medical Genetics Unit, Hospital Pediátrico, Centro Hospitalar e Universitário de Coimbra, Coimbra, Portugal and University Clinic of Pediatrics, Faculty of Medicine, University of Coimbra, Portugal*

*aluisa.dcarvalho@gmail.com*

**Introduction**: Frontotemporal dementia is the second most frequent form of early-onset dementia. Its molecular basis is heterogeneous. Heterozygous mutation in the progranulin gene (*GRN*) is a frequent cause of this disease, with autossomal dominant inheritance. Recently, Smith *et al*. presented two brothers with adult-onset neuronal ceroid lipofuscinosis and homozygous mutation in the *GRN* gene (c.813_816del (p.Thr272Serfs*10)). This type of neuronal ceroid lipofuscinosis was designated type 11 (CLN11).

**Case report**: We report one family with frontotemporal dementia with molecular diagnosis: heterozygous for g.22632264dupGT (p.Ser301Cysfs*60) mutation in the *GRN* gene. One member of this family presented with progressive visual failure at 25 years, followed by dystonia with muscle weakness, multifocal myoclonus and dysarthria. Plasma progranulin values are undetectable. The molecular analysis of *GRN* gene revealed a homozygous mutation g.22632264dupGT (p.Ser301Cysfs*60) confirming the diagnosis of CLN11.

**Comment**: Mutations in specific genes usually determine important phenotypes in either the heterozygous or homozygous state. In this two families, with mutations in the *GRN* gene, two clinical distinct neurological disorders are present: frontotemporal dementia at heterozygous and CLN11 at homozygous state. A deletion was present in Smith *et al*. previously reported family. The family reported by the authors is the first with homozygous state for a duplication in the *GRN* gene.