molecular testing of *DCX* is warranted in the mothers of affected children, allowing genetic counselling to at-risk family members, including prenatal diagnosis and preimplantation genetic diagnosis. Massive parallel sequencing, either whole exome or gene panels, of females with ID with or without epilepsy will likely increase detection of mutations in the *DCX* gene.

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A PORTUGUESE FAMILY WITH CADASIL DIAGNOSIS WITH ANTICIPATION AGE OF ONSET OBSERVED

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Introdution: Cerebral Autossomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (CADASIL) is a rare hereditary disease characterized by recurrent transient ischemic attacks, strokes, and vascular dementia.

Since it is a dominant disease, heterozygous and homozygous patients are expected to be clinically indistinguishable. Nevertheless, some homozygous patients with CADASIL have been reported and in some cases with a severe phenotype.

Case report: We would like to report a Portuguese family with inbreeding with diagnosis of CADASIL. It has been found the p.Arg558Cys mutation in *NOTCH3* gene in six members of this family studied, two of them in homozygosity. One of the homozygous case present a more severe phenotype compared with his relatives with an age of onset at 10 years old. According to this finding, we wonder if the homozygosity can justify this early age of onset case or its severity.

Discussion: Differences in clinical profile between homozygous and heterozygous of this family members and between other CADASIL families with homozygosity described should be discuss in order to understand if the homozygosity state increases the pathologic consequences of the mutation providing a more severe and early phenotype.