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Cerebral cavernous malformations, CCM genetic test and follow-up of patients and their relatives: Our data

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Cerebral Cavernous Malformations (CCM) are vascular lesions involving brain capillaries. CCM is considered a rare genetic disorder however, its incidence is underestimated. Many cases, instead, are post-mortem accidentally detected, being asymptomatic about 30% of affected. Most common clinical manifestations include intracerebral hemorrhage, seizures, focal neurological deficits. Resonance Magnetic Image (RMI) performed with "gradient-echo" sequences is the most effective diagnostic method. CCM can arise sporadically or be inherited as autosomal dominant character leading to onset of familiar forms. Mutations at the three loci CCM1/KRIT1, CCM27MGC4607, CCM3/PDCD10 were detected in about 90% and 55% of patients affected by familial and sporadic forms, respectively. In the last 10 years, more than 100 CCM cases arrived to our laboratory for molecular diagnosis. Our diagnostic procedure includes both direct sequencing and Multiplex Ligation-Dependent Probe Amplification (MLPA) performed for the three CCM genes on DNA sample extracted from peripheral blood, in order to detect point mutations or large genomic rearrangement, respectively. About mutations frequencies, our data overlap with literature's ones. However, we noticed recurrent polymorphisms overall in CCM1 and CCM2 genes. Several case-control studies were performed and rs17164451, rs11542682 in CCM1 and rs11552377, rs73107990, rs2289367 in CCM2 resulted associated with an increased risk of sporadic CCM development. Moreover, several were detected in patients showing more severe symptomatology. Therefore, our aim is to integrate anamnesis and RMI data with genetic test results in order to establish possible genotype-phenotype correlations usable both as follow-up route for patients and as prognostic factors for their relatives.

Biography

Antonina Sidoti was born in Messina, Italy. She obtained her degree in "Biological Sciences" (1992), magna cum laude, at the University of Messina, Italy. In 1993 she obtained professional qualification and registration in the National Board of Biologists. Following a four years specialization course, she obtained in 1997 at the University of Messina, the specialization degree in"Clinical Pathology". In 1998 she was winner of a foreign study scholarship held in Paris at the Biology Lab Molecular and Genetic Prof. Luc Montagnier, "Center Integré de RecherchesBiocliniques sur le Sida ".In 1999 she obtained PhD in "Cellular Biology and Genetics", University of Messina. In 1999-2000 she partecipated to a selection for University Researcher at the Medical School of the University of Messina: she won the selection and became University Researcher, in the field of Applicated Biology (BIO/13). Currently sheis Associated Professor in Applied Biology and Chairman of master's degree course in "Biotechnology for the Health" of University of Messina, Italy. She plays her teaching activity in Degree in Medicine, Biotechnology and other degrees of same University. Actually her main research field is related to molecular and genetics features of Cerebral Cavernous Malformations; identification of candidate genes and causative variants in different forms of Retinitis Pigmentosa and molecular genetic features and genotype-phenotype correlations in Trimethylaminuria.

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