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P38. CORPUS CALLOSUM ABNORMALITIES AND AUTISM SPECTRUM DISORDERS

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The corpus callosum (CC) represents the most important portion of white matter in the human brain. Corpus callosum abnormalities (CCA) have an estimated prevalence ranging from 0.3% up to 0.7% in patients undergoing brain imaging. Furthermore, alterations of the white matter are associated with dysfunctional cognitive and social abilities. Recent studies suggest the hypothesis of a correlation between CCA and autism spectrum disorders (ASD). We reviewed MRI data, clinical and genetics findings of patients with ASD recruited in the series of the Italian CCA Study Group. We found 26/579 individuals with ASD (10%), 8 females and 18 males. Complete CC agenesis was present in 3/26 cases (12%), partial agenesis in 9/26 (35%), thin CC in 4/26 (15%), thick CC in 10/26 (38%). Non-syndromic CCA ASD subjects were 18/26 (69%). Intellectual disability was referred in 25/26 patients (96%) with prevalent mildmoderate degree (62%). Chromosomal micro array (CMA) data were available in 22/26 (87%). CMA was negative in 15/22 (68%) and variants of unknown significance were reported in 7/22 (32%). Whole exome sequencing (WES) is in progress in a few cases. These data support the hypothesis that CCA constitutes a major risk factor for developing ASD with low cognitive functions. Individuals with CCA should be screened for ASD and CCA should be considered in autism diagnostic evaluations as well. Combined CMA and Next-generation sequencing (NGS) strategies will increase the probability to identify new causative genes for CCA and ASD. Further studies are necessary to define the role of CCA in ASD.