Disclosure

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Weak evidence for a relation between bipolar disorder and heterozygous *ZNF92* and *CLN6* variants

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We read with interest the article by Privitera et al.[1] about a study of a five-generation family in which 12 members have bipolar disorder, eight of whom underwent whole exome sequencing to detect a common underlying genetic defect. Three patients who underwent whole exome sequencing had bipolar disorder and one was a "borderline" case.[1] It was found that a heterozygous missense variant in *CLN6* was associated with the "borderline" phenotype and the combination of heterozygous missense variants in *CLN6* and *ZNF92* was associated with the bipolar phenotype.[1] The study is appealing but raises concerns that should be discussed.

We disagree with the conclusions that the "borderline" case was due to the heterozygous *CLN6* variant and that the bipolar disorder was due to the combination of the heterozygous *ZNF92* variant and the heterozygous *CLN6* variant.[1] No studies were conducted to confirm that either the *CLN6* or *ZNF92* gene products were dysfunctional. Furthermore, previous studies have shown that heterozygous variants in either gene are not pathogenic. Only homozygous or compound heterozygous *CLN6* or *ZNF92* variants have been found pathogenic and were associated with depression and anxiety.[2,3]

One limitation of the study is its small sample. To demonstrate an effect of *CLN6* and *ZNF92* variants on psychological perception, larger cohorts with bipolar disorder are needed.

Another argument against bipolar disorder as a phenotypic manifestation of *CLN6* and *ZNF92* variants is that none of the mutation carriers manifested with phenotypic features of neuronal ceroid lipofuscinosis (NCLs) other than psychiatric disease. NCLs are a heterogeneous group of neurodegenerative diseases, characterized by progressive cerebral atrophy due to lysosomal storage. Common clinical features include epileptic seizures, progressive cognitive and motor decline, and visual impairment, which occur over different time points according to the subtype.[4] The main clinical features include progressive deterioration of cognitive functions and pigmentary retinal degeneration.[5] In some of these patients, dementia is associated with personality and behavior changes, suggesting a psychotic disorder with dysarthria and tic-like dyskinetic movements.[5] NCLs may have juvenile or adult onset. Adult NCL is also known as Kufs disease.[5] Patients with juvenile NCL often have severe psychiatric symptoms. These are common in the mid-teens and include symptoms such as anxiety and affective and psychotic disorders. This is why mutation carriers should have undergone investigations with cerebral magnetic resonance imaging, electroencephalography, and of the cerebrospinal fluid.

Since NCLs frequently manifest with pigmentary retinal degeneration, readers should be informed whether any of the eight investigated patients were visually impaired. Moreover, there were also no neuropsychological investigations to determine the presence of cognitive impairment.

Overall, this interesting study has limitations that call both the results and their interpretation into question. Clarifying these weaknesses would strengthen the conclusions and could improve the study.

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