No evidence for an involvement of copy number variation in *ABCA13* in schizophrenia, bipolar disorder, or major depressive disorder

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Previously, Knight and colleagues reported a patient with schizophrenia (SCZ) who carried the complex chromosomal rearrangement inv(7)(p12.3;q21.11), t(7;8)(p12.3;p23). The translocation between chromosomes 7 and 8 resulted in a disruption of *ABCA13*, suggesting a possible role for this gene in the etiology of SCZ. This gene is member 13 of the ATP-binding cassette transporter subfamily A. Protein expression analyses in the adult mouse showed Abca13 expression in the brain (Knight *et al.*, 2009).

Subsequent resequencing of functional domain exons identified rare variants that were associated with both SCZ and bipolar disorder (BPD; Knight *et al.*, 2009). Furthermore, the same research group screened an SCZ and BPD sample for the presence of small copy number variants (CNVs) in this gene. Multiplex amplicon quantification assays were performed on DNA samples from 1004 patients with SCZ, on 428 patients with BPD, and on 1086 controls, and no over-representation of CNVs among patients compared with controls was identified (Pickard *et al.*, 2012).

We sought to elucidate further the potential role of CNVs in *ABCA13* and analyzed three clinically well-characterized data sets for the occurrence of CNVs in *ABCA13*: (i) 575 patients with major depressive disorder (MDD; Degenhardt *et al.*, 2012), (ii) 882 patients with BPD (Priebe *et al.*, 2012), and (iii) 1637 patients with SCZ or schizoaffective disorder. Of them, 487 patients with SCZ have previously been included in a GWA study by Rietschel *et al.* (2011). In addition, 1618 controls (described in detail in Degenhardt *et al.*, 2012) were screened for the presence of CNVs in *ABCA13*. All participants were of German descent according to self-reported ancestry, and each participant provided written informed consent before inclusion.

All individuals were genotyped on HumanHap550v3, Human610-Quadv1, and Human660W-Quad arrays (Illumina, San Diego, California, USA). Detailed information on CNV detection and CNV quality control is provided elsewhere (Degenhardt *et al.*, 2012). Only those markers common to all three chips were analyzed. Participants were excluded if their SD from the log *R* ratio calculated over all SNPs exceeded 0.30.

The three data sets were analyzed for the presence of both microdeletions and microduplications including the *ABCA13* gene plus 10 kb upstream and downstream of the Refseq gene boundary (chr7: 48208389–48657637; hg18). All CNVs were required to span a minimum of 10 consecutive SNPs and have a log Bayes Factor (QuantiSNP) or confidence value (PennCNV) of at least 10.

No CNV including *ABCA13* was identified in our three patient samples, nor in the controls. Hence, we did not find any evidence for CNVs in *ABCA13* as a risk factor for the development of SCZ, BPD, or MDD. It cannot be excluded, however, that extremely rare CNVs might play a role in disease development, which would only be detectable in considerably larger samples. It is therefore important to gather data from several large studies before final conclusions regarding the involvement of CNVs in *ABCA13* in SCZ, BPD, or MDD can be drawn.

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Conflicts of interest

There are no conflicts of interest.

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