Single nucleotide variations encoding missense mutations in G protein-coupled receptors may contribute to autism.

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Abstract

Autism is a neurodevelopmental condition with a range of symptoms that vary in intensity and severity from person to person. Genetic sequencing has identified thousands of genes containing mutations in autistic individuals, which may contribute to the development of autistic symptoms. Several of these genes encode G protein-coupled receptors (GPCRs) which are cell surface expressed proteins that transduce extracellular messages to the intracellular space. Mutations in GPCRs can impact their function, resulting in aberrant signalling within cells, and across neurotransmitter systems in the brain. This review summarises the current knowledge on autism-associated single nucleotide variations encoding missense mutations in GPCRs, and the impact of these genetic mutations on GPCR function. For some autism-associated mutations, changes in GPCR expression levels, ligand affinity, potency and efficacy have been observed; however, for many the functional consequences remain unknown. Thus, further work to characterise the functional impacts of the genetically identified mutations is required.

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FIGURE 1

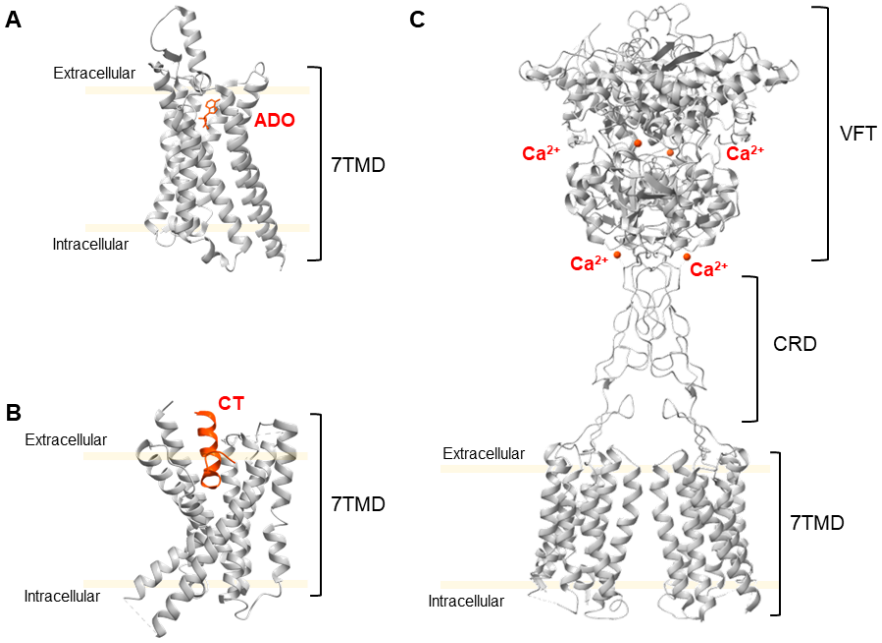

FIGURE 2