

# Schizophrenia

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## Characteristics

Schizophrenia is a psychiatric disorder in which individuals have periods of psychosis. Schizophrenia is characterized by positive and negative symptoms. Positive symptoms include hallucinations (a sensory perception in the absence of a stimulus), delusions (a fixed, unshakable belief that is false), and disorganization of thought (e.g., making up new words) and speech (e.g., tangentiality). Negative symptoms include a flat mood, lack of energy, poverty of speech, and apathy.

Other symptoms include impairments in cognition, such as problems with memory, paying attention, and carrying out executive functions. Also mood disorders and anxiety are common.

Multiple genetic risk factors have been identified in linkage analysis studies of individuals with schizophrenia, including variants in the following genes: *APOE*, *COMT*, *DAO*, *DRD1*, *DRD2*, *DRD4*, *DTNBP1*, *GABRB2*, *GRIN2B*, *HP*, *IL1B*, *MTHFR*, *PLXNA2*, *SLC6A4*, *TP53*, and *TPH1* (1).

Microdeletions and microduplications have been found to be three times more common in individuals with schizophrenia, compared to controls. Because these deletions and duplications are in genes that are overexpressed in pathways related to brain development, it is possible that the inheritance of multiple rare variants contributes to the development of schizophrenia (2).

## Diagnosis

The diagnosis of schizophrenia is made via a psychiatric assessment using the criteria presented in the American Psychiatric Association Manual of Psychiatric Diseases, 4th edition (DSM-IV). To make a diagnosis, specific characteristic symptoms of schizophrenia must be present for at least 6 months, together with a disruption in social or occupational function, in the absence of another diagnosis that could account for the symptoms.

Although schizophrenia is highly heritable, the genetics are complex and molecular genetic testing is not currently available.

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## Management

*Treatment of manifestations:* Antipsychotic medications are the mainstay of treatment and are effective in reducing symptoms and improving behaviors in patients with schizophrenia, although the type, dose, and route of administration of antipsychotics depends upon the clinical scenario. Adverse effects are common, and may require the dose or type of drug to be altered. Antipsychotics may be given with counseling and other types of psychosocial interventions. For refractory symptoms, an alternative antipsychotic or an additional antipsychotic may be required.

*Surveillance:* Routine monitoring for the symptoms and signs of extrapyramidal adverse effects is needed in individuals taking antipsychotics. These adverse effects include akathisia (feeling of restlessness that may be accompanied with motor restlessness), dystonias (involuntary contraction of large muscle groups), and parkinsonian syndrome. Patients should also be monitored for signs of tardive dyskinesia (involuntary facial movements) and drug-specific adverse effects.

*Prevention of secondary complications:* Patients should be regularly monitored for weight gain and metabolic problems such as hyperglycemia and hyperlipidemia.

## Genetic Counseling

Genetic counseling is recommended for people who have a family member with schizophrenia. Recurrence risk counseling is based on empiric familial risk for families with individuals with schizophrenia (3).

The lifetime risk of schizophrenia for the general population is estimated to be 0.2 to 0.7% (4). In siblings of patients, the risk is 10%, and in the children of patients, 13%. The risk for second-degree relatives is 5% (3).

## References

1. Allen N.C., Bagade S., McQueen M.B., Ioannidis J.P. et al. *Systematic meta-analyses and field synopsis of genetic association studies in schizophrenia: the SzGene database.* Nature genetics. 2008;40(7):827–34. PubMed PMID: 18583979.
2. Walsh T., McClellan J.M., McCarthy S.E., Addington A.M. et al. *Rare structural variants disrupt multiple genes in neurodevelopmental pathways in schizophrenia.* Science. 2008;320(5875):539–43. PubMed PMID: 18369103.
3. Papadimitriou G.N., Dikeos D.G. *How does recent knowledge on the heredity of schizophrenia affect genetic counseling?* Current psychiatry reports. 2003;5(4):239–40. PubMed PMID: 12857525.
4. Kendler K.S., Gallagher T.J., Abelson J.M., Kessler R.C. *Lifetime prevalence, demographic risk factors, and diagnostic validity of nonaffective psychosis as assessed in a US community sample. The National Comorbidity Survey.* Archives of general psychiatry. 1996;53(11):1022–31. PubMed PMID: 8911225.

## Tests in GTR by Condition

Schizophrenia