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Choose a disease of your interest and investigate about it using BOTH Open Targets and HumanMine platforms.

Write what you have learned about it and upload a report here (max 4-5 pages, you can include screenshots).

You can do this in groups of 2-3, but everyone needs to submit their own report. If you decide to do it in groups please include the names of all the group members at the beginning of the report.

SCHIZOPHRENIA

Genetic association:

There is low genetic association overall, but the highest associations scores are using open target genetics and UniProt Literature

Somatic mutations:

There are no somatic mutations on any target

Drugs:

ChEMBL has a very high score in most targets

Affected pathways:

No targets are affected by pathway analysis

The RNA expression and animal models have almost no target score, the animal models having a little higher score.



Associated targets:

First of all, we can observe it has around 5774 different entries. It has a really high overall association score, with at least 500 entries with a high score.

It has no somatic mutations, also no pathways and systems biology. And it has a really poor RNA expression, with almost no entries, and the ones there have really low scores.

It is important to remark that it has lots of drugs associated with it, and almost all of them with the maximum score possible.

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Symbol	Overall association score	Genetic associations	Somatic mutations	Drugs	Pathways & systems biology	Text mining	RNA expression	Animal models	Target name
DRD2									dopamine receptor D2
DRD3									dopamine receptor D3
GRIN2A									glutamate ionotropic receptor NMDA type subunit 2A
ESR2									estrogen receptor 2
RTN4R									reticulin 4 receptor
MTHFR									methylenetetrahydrofolate reductase
SHANK3									SH3 and multiple ankyrin repeat domains 3
HTR1A									5-hydroxytryptamine receptor 1A
HTR2A									5-hydroxytryptamine receptor 2A
DRD4									dopamine receptor D4
SLC6A4									solute carrier family 6 member 4
HTR2C									5-hydroxytryptamine receptor 2C
SLC6A3									solute carrier family 6 member 3
HTR4									5-hydroxytryptamine receptor 4
HTR6									5-hydroxytryptamine receptor 6
HTR1B									5-hydroxytryptamine receptor 1B
NDUFAF2									NADH:ubiquinone oxidoreductase complex assembly factor 2
HTR3A									5-hydroxytryptamine receptor 3A
HTR7									5-hydroxytryptamine receptor 7
HTR5A									5-hydroxytryptamine receptor 5A
HTR3B									5-hydroxytryptamine receptor 3B
GABRA5									gamma-aminobutyric acid type A receptor subunit alpha5
SLC6A2									solute carrier family 6 member 2
HTR3C									5-hydroxytryptamine receptor 3C
HTR3D									5-hydroxytryptamine receptor 3D
HTR3E									5-hydroxytryptamine receptor 3E
HTR1D									5-hydroxytryptamine receptor 1D
HTR1F									5-hydroxytryptamine receptor 1F
HTR2B									5-hydroxytryptamine receptor 2B
NDUFA10									NADH:ubiquinone oxidoreductase subunit A10

Profile:

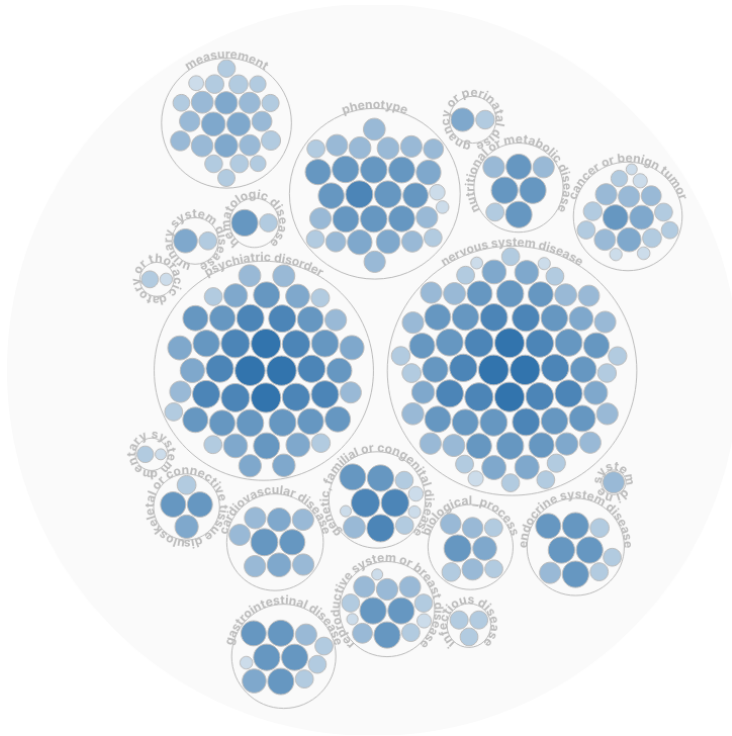
These are known drugs that affect schizophrenia through different mechanisms

Disease information	Drug information				Target information	C
Disease	Drug	Type	Mechanism Of Action	Action Type	Symbol	n
schizophrenia	ACAMPROSATE	Small molecule	Glutamate [NMDA] receptor antagonist	Antagonist	GRIN2C	g
schizophrenia	CHLORPROMAZINE	Small molecule	D2-like dopamine receptor antagonist	Antagonist	DRD4	d
schizophrenia	AMISULPRIDE	Small molecule	Serotonin (5-HT) receptor antagonist	Antagonist	HTR1B	5
schizophrenia	KETAMINE	Small molecule	Glutamate [NMDA] receptor negative allosteric modulator	Negative allosteric modulator	GRIN2A	g
schizophrenia	METFORMIN	Small molecule	Mitochondrial complex I (NADH dehydrogenase) inhibitor	Inhibitor	MT-ND6	n
schizophrenia	AMISULPRIDE	Small molecule	Serotonin (5-HT) receptor antagonist	Antagonist	HTR3D	5
schizophrenia	CHLORPROMAZINE	Small molecule	D2-like dopamine receptor antagonist	Antagonist	DRD3	d
schizophrenia	METFORMIN	Small molecule	Mitochondrial complex I (NADH dehydrogenase) inhibitor	Inhibitor	MT-ND4	n
schizophrenia	ZIPRASIDONE	Small molecule	Serotonin 2c (5-HT2c) receptor antagonist	Antagonist	HTR2C	5
schizophrenia	AMISULPRIDE	Small molecule	Serotonin (5-HT) receptor antagonist	Antagonist	HTR2A	5

Rows per page: 10 1-10 of 2137

We can look at each target and its relations individually

For example for the gene of dopamine receptor D2 (DRD2) Open Target can associate 758 diseases or phenotypes:



We can even filter the associations to this gene by evidence, disease and therapeutic areas

Evidence-specific filters

Data Types

Disease/phenotype-specific filters

Therapeutic Areas

☐ Biological_proce... 18

☐ Cancer or benign... 106

☐ Cardiovascular ... 42

☐ Disorder of ear 1




☐ Disorder of visu... 38

☐ Endocrine syste... 143







In the HumanMine platform we can see alleles related to the disease, for example:

Alleles (77)	
Showing 1 to 5 of 77 rows	<div> ^ OPTIONS </div> <div> Rows per page: 5 </div> <div> Page 1 </div>
<div> 🔍 ⌵ 📄 </div> Alleles Primary Identifier	<div> 🔍 ⌵ 📄 </div> Organism Name
1320988	Homo sapiens
153362	Homo sapiens
207488	Homo sapiens
226431	Homo sapiens
226432	Homo sapiens

We can also see datasets that present information about this disease, like OMIM-diseases, clinvar and hpo-annotation

Data Sets (3)			
Showing 1 to 3 of 3 rows		^ OPTIONS	Rows per page: All (3) < > Page 1 < >
 Data Sets Name	 Data Sets URL	 Data Source Name	
OMIM diseases	NO VALUE	OMIM	
clinvar	https://www.ncbi.nlm.nih.gov/clinvar/	NCBI	
hpo-annotation	NO VALUE	Human Phenotype Ontology	

This website also shows us genes related to this database

Genes (11)						
Showing 1 to 11 of 11 rows		^ OPTIONS	Rows per page: All (11) < > Page 1 < >			
 Genes Symbol	 Genes Name	 Genes Primary Identifier	 Genes Secondary Identifier	 Genes Length	 Organism Name	
APOL2	apolipoprotein L2	23780	ENSG00000128335	13746	Homo sapiens	
APOL4	apolipoprotein L4	80832	ENSG00000100336	15706	Homo sapiens	
CHI3L1	chitinase 3 like 1	1116	ENSG00000133048	7774	Homo sapiens	
COMT	catechol-O-methyltransferase	1312	ENSG00000093010	28204	Homo sapiens	
DAOA	D-amino acid oxidase activator	267012	ENSG00000182346	25168	Homo sapiens	
DISC2	disrupted in schizophrenia 2	27184	NO VALUE	3892	Homo sapiens	
DRD3	dopamine receptor D3	1814	ENSG00000151577	71828	Homo sapiens	
HTR2A	5-hydroxytryptamine receptor 2A	3356	ENSG00000102468	66537	Homo sapiens	
MTHFR	methylenetetrahydrofolate reductase	4524	ENSG00000177000	20381	Homo sapiens	
RTN4R	reticulon 4 receptor	65078	ENSG00000040608	26904	Homo sapiens	
SYN2	synapsin II	6854	ENSG00000157152	187667	Homo sapiens	