TOOLBOX

New resource compiles data on autism-linked genes

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A new online database provides searchable information for nearly 10,000 genes, variants and chromosomal regions linked to autism. Researchers describe the resource, dubbed **AutismKB**, in the January issue of *Nucleic Acids Research*[1].

Over the past few years, numerous mutations and genetic variants have been linked to autism. Most cases of autism probably result from carrying **a combination of these variants**.

According to the researchers, there are three existing databases, including **SFARIGene** — funded by the Simons Foundation, SFARI.org's parent organization — that summarize and **rank the evidence** linking various genes to autism.

The new database includes autism candidate genes from a wider range of studies than do the other resources, the researchers say.

They searched the abstracts of more than 4,000 articles and identified 11 genome-wide association studies, 242 association studies that focus on candidate genes, 13 studies of gene expression, 95 studies of **copy number variants** — duplications or deletions of DNA — 23 genome-wide analyses that associate variants across a chromosomal region and 236 studies that link a particular gene or protein to autism.

They also searched the **Online Mendelian Inheritance in Man**, or OMIM, database for autism-related disorders such as **fragile X** and Rett syndromes.

The researchers **ranked the genes** from these studies by assigning a number to each based on the strength of the evidence.

For example, for diseases associated with autism symptoms in a single individual, genes receive a score of one, whereas for diseases that show autism symptoms in multiple individuals and studies, they receive a score of four. The researchers then added up the scores for each study that links a gene to autism.

The resource is a comprehensive overview of the literature, but may be too inclusive, some experts caution. For example, it includes genome-wide association studies with few participants, which many researchers argue are often statistically weak.

However, users of the database can manually rank what they perceive to be the significance of various approaches. For example, they can assign a score of one to low-scale association studies.

The database also includes functional information about each gene, collected from various sources, such as the **Allen Brain Atlas** and the **Mouse Genome Informatics** resource. The functional pathways most highly associated with autism are those that involve **synapses**, the junctions between neurons, the study found.

AutismKB also includes clinical information from each study. For example, it lists participants' ancestral background, age and whether their autism was diagnosed using the Autism Diagnostic Observation Schedule or the Autism Diagnostic Interview-Revised, two widely used diagnostic tests.

References:

1: Xu L.M. *et al. Nucleic Acids Res.* **40**, D1016-1022 (2012) **PubMed**