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GENETIC DATABASE WEBSITE

HEALTHCARE IT

Database that consolidates all disease variants observed and reported in scientific literature, specifically for de novo mutations.

TECHNOLOGY TYPE

Software
Disease
Genetics

STAGE OF DEVELOPMENT

- Over 160 publications extracted and verified.

- Information from additional publications still needs to be extracted.

LEARN MORE

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TECHNOLOGY SUMMARY

The most common method of identifying de novo mutations involves searching research databases, such as PubMed, for specific genes. With almost 15,000 articles related to de novo mutations, identifying particular genes and their associated diseases is time consuming.

The Genetic Database Website provides access to curated information regarding all de novo mutations observed and reported in scientific literature. Using the database, researchers can check whether a de novo mutation in a gene or a more specific locus has been reported previously. Those investigating rare diseases will be able to eliminate some findings quickly as non-causally related if that mutation is observed across other, more common diseases in the database.

FEATURES AND BENEFITS

- Increases clinician and researcher access to genetic disease information.
- Facilitates rapid identification of diseases related to specific de novo mutations.

INVENTOR PROFILE

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