

Projeto MC536

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Banco de Dados: <https://www.uniprot.org/diseases/>

- Banco de doenças;
- Nome das doenças, categoria e descrição;

The screenshot shows the UniProt Human diseases results page. At the top, there is a search bar with 'Human diseases' selected. Below the search bar, the page title 'Human diseases results' is displayed. A text box explains that human diseases in UniProtKB entries are described in a controlled vocabulary and provides a link to the user manual. Below this, a 'MapTo' section shows a list of diseases. The first disease listed is '2,4-dienoyl-CoA reductase deficiency', which is described as a rare, autosomal recessive, inborn error of polyunsaturated fatty acids and lysine metabolism. The second disease listed is '2-aminoacidic 2-oxoadipic aciduria', described as a metabolic disorder characterized by increased levels of 2-oxoadipate and 2-hydroxyadipate in the urine and plasma. At the bottom, a blue banner contains a privacy notice regarding the GDPR.

UniProt

Human diseases

BLAST Align Retrieve/ID mapping Peptide search

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Human diseases results

The human diseases in which proteins are involved are described in UniProtKB entries with a controlled vocabulary. Information about the usage of this controlled vocabulary in UniProtKB entries can be found in the user manual.

By default, searching the diseases will look for matches in both name and description. Example queries to search diseases only by name: name:disorder - name:syndrome

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MapTo

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Disease
<input type="checkbox"/> 2,4-dienoyl-CoA reductase deficiency A rare, autosomal recessive, inborn error of polyunsaturated fatty acids and lysine metabolism, resulting in mitochondrial dysfunction. Affected individuals have a severe encephalopathy with neurologic and metabolic abnormalities beginning in early infancy. Laboratory studies show increased C10:2 carnitine levels and hyperlysinemia. UniProtKB (3)
<input type="checkbox"/> 2-aminoacidic 2-oxoadipic aciduria A metabolic disorder characterized by increased levels of 2-oxoadipate and 2-hydroxyadipate in the urine, and elevated 2-aminoadipate in the plasma. Patients can have mild to severe intellectual disability, muscular hypotonia, developmental delay, ataxia, and epilepsy. Most cases are asymptomatic. UniProtKB (1)

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Análises

- Suportes

- Confianças



Predição



Exemplo

	Síndrome	Autossomo Dominante	Mental
Síndrome	1	0.008	0,023
Autossomo Dominante	0,045	1	0,005
Mental	0,163	0,006	1