Superoxide Dismutase 1 (SOD1)

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1 Overview

Superoxide Dismutase 1 (SOD1) refers to a specific gene on chromosome 21[1].

2 Function

2.1 Prevention of Neuronal Degeneration

SOD1 encodes for enzymes such as CuZnSOD. CuZnSOD along with the other SOD isoforms play a role in removing oxygen free radicals from the body[1]. Gene mutations lead to less activity of CuZnSOD, resulting in accumulation of free radicals and resulting in neuronal degeneration[1].

Most mutations in FALS have shown only modest reductions in enzyme activity[1]. Leading researchers to believe that a mutant SOD1 protein may have actively toxic properties[1].

3 Mutation

3.1 Amyotrophic Lateral sclerosis

- One of 100s of mutations in SOD1 have been linked to hereditary ALS[1].
- Studies of patients with adult-onset FALS have determined that about 20% of these individuals have mutations in SOD1[1]

1. O'Sullivan SB, Schmitz TJ, Fulk GD, eds. *Physical Rehabilitation*. 7th ed. F.A. Davis Company; 2019.