

Adenosine Deaminase Deficiency

Adenosine deaminase deficiency is an autosomal recessive metabolic disorder that is one of the major causes of severe combined immunodeficiency (SCID). Adenosine deaminase is an enzyme involved in purine metabolism. It is used to break down adenosine to inosine for the turnover of nucleic acids for DNA synthesis and cell growth. This deficiency leads to an accumulation of deoxyadenosine which leads to a buildup of ATP. High ATP, which normally activates RNR, leads to high dATP, which inactivates the enzyme ribonucleotide reductase, which is a key enzyme in DNA synthesis. This in turn prevents DNA synthesis. Because developing T cells and B cells are extremely mitotically active, these cells are susceptible to cell death. Loss of B and T cell function causes severe immunodeficiency.



PLAY PICMONIC

Adenosine to Inosine

[A-dentist-singing becoming N'SYNC](#)

Adenosine deaminase is an enzyme involved in purine metabolism. It is used to breakdown adenosine to inosine for the turnover of nucleic acids for DNA synthesis and cell growth.

Excess ATP

[Excess ATP-batteries](#)

Adenosine deaminase deficiency leads to an accumulation of deoxyadenosine which leads to a buildup of ATP and dATP in cells.

Inhibition of Ribonucleotide Reductase

[Red-bull Red-duck](#)

High ATP, which normally activates RNR, leads to high dATP, which inactivates the enzyme ribonucleotide reductase, which is a key enzyme in DNA synthesis.

Prevents DNA Synthesis

[DNA Double-helix](#)

High ATP, which normally activates RNR, leads to high dATP. This inactivates the enzyme ribonucleotide reductase, which is a key enzyme in DNA synthesis. Thus, DNA synthesis is prevented.

Decreases B Cells and T Cells

[Down-arrows on Basketball and Tennis-ball](#)

Developing B cells and T cells are extremely mitotically active and thus largely affected by the disruption in DNA synthesis. Lack of B and T cells results in high susceptibility to viral, fungal and bacterial infections.

Major cause of SCID

[SCID marks](#)

Adenosine deaminase deficiency accounts for about 15% of cases of severe combined immunodeficiency (SCID).