

# SEVERE COMBINED IMMUNODEFICIENCY

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## SEVERE COMBINED IMMUNODEFICIENCY: RELEVANT FACTS

- deoxynucleotides (Figure 2)
  - ✓ ribonucleotide reductase acts on nucleotides (i.e., ATP, GTP, CTP, UTP) to form their deoxynucleotide forms (i.e., dATP, dGTP, dCTP, dUTP {also called TTP})
  - ✓ all four are essential for the formation of DNA
  - ✓ dATP inhibits ribonucleotide reductase to block formation of dGTP, dCTP and TTP
  - ✓ diminished production of the deoxynucleotides prevents maturation of newly formed cells of the immune system to functional cells
- methylation reactions (Figure 3)
  - ✓ S-adenosylmethionine provides the methyl group for these reactions producing S-adenosylhomocysteine in the process
  - ✓ important for cell growth
  - ✓ excess S-adenosylhomocysteine is toxic to immature cells of the immune system by blocking the methylation reactions and preventing them from becoming functional cells
- adenosine deaminase deficiency causes accumulation of deoxyadenosine:
  - ✓ deoxyadenosine converted to excess dATP that inhibits ribonucleotide reductase (see above)
  - ✓ deoxyadenosine blocks processing of S-adenosylmethionine that accumulates to block methylation reactions (see above)



