

CARTILAGE-HAIR HYPOPLASIA

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- defective in RMRP gene produces a unique form of RNA that does not provide coding information
- unique RNA attaches to several proteins to form an enzyme, mitochondrial RNA-processing endoribonuclease (RNase MRP)
- RNase MRP functions seem to include:
 - helping to make copies of DNA in mitochondria, the energy-producing center of the cell
 - processing of ribosomal RNA that is required in the assembly of amino acids into proteins
 - control of the cell cycle needed for the cell to replicate (duplicate) itself
- mutations of RMRP create an unstable molecule that is then unable to create the RNase MRP
- consequences lack of RNase MRP are indicated in the figure

