APRT (adenine phosphoribosyltransferase) is a 180 amino acid protein that localizes to the cytoplasm and belongs to the purine/pyrimidine phosphoribosyltransferase family. Existing as a homodimer, APRT functions to catalyze the formation of inorganic pyrophosphate and AMP from adenine and 5-phosphoribosyl-1-pyrophosphate (PRPP), a reaction that is essential for both purine metabolism and AMP biosynthesis. Defects in the gene encoding APRT are the cause of APRT deficiency, also known as 2,8-dihydroxyadenine urolithiasis, which is an autosomal recessive disease that results in renal failure. The gene encoding APRT maps to human chromosome 16, which encodes over 900 genes and comprises nearly 3% of the human genome. The GAN gene is located on chromosome 16 and, with mutation, may lead to giant axonal neuropathy, a nervous system disorder characterized by increasing malfunction with growth. The rare disorder Rubinstein-Taybi syndrome is also associated with chromosome 16, as is Crohn’s disease, which is a gastrointestinal inflammatory condition.

REFERENCES