Climb™

Supporting those affected by Inherited Metabolic Disorders

Adenine Phosphoribosyltransferase Deficiency

Adenine Phosphoribosyltransferase (APRT) Deficiency is a rare disorder that is caused by a lack of APRT, an enzyme which is involved in converting a building block of DNA known as adenine into a molecule called Adenosine Monophosphate (AMP) when it is needed for cell energy. Because of the enzyme deficiency the Adenine is converted to an insoluble substance which crystallises and forms stones in the kidneys and urinary tract. Recurring stones are the main symptom of the disorder, causing blockages and resulting pain and difficulty passing urine. Untreated this can cause severe kidney problems. Severity of APRT Deficiency is extremely variable.

Synonyms

Alternative names for this condition are:

- 2,8-dihydroxyadenine urolithiasis
- 2,8-dihydroxyadeninuria
- APRT deficiency
- DHA crystalline nephropathy

Further information about this condition is available from Climb.

Disclaimer

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