Phosphoribosyl Pyrophosphate Synthetase Deficiency

Other names that may be used for this disorder are:
- PRPP Synthetase Deficiency
- PRPS Deficiency

Phosphoribosyl Pyrophosphate Synthetase (PRPS) Deficiency is a rare purine disorder. A purine is any of a group of organic compounds that are found in tissues. The two major purines are adenine and guanine. Others include uric acid, xanthine and hypoxanthine. This disorder causes convulsions, a low number of red blood cells (anaemia), autistic-like behaviour and severely impaired intelligence and social ability. Individuals also have a delay in reaching developmental milestones such as speech.

Treatment for individuals with this disorder aims to provide relief for any symptoms and support in the care of the individual.

This information is fully sourced and referenced, for more detailed information and references please contact CLIMB by email, letter or telephone.

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