Hyperprolinaemia Type 2

Other names for this condition are:
- 1-@ Pyrroline-5-Carboxylate Dehydrogenase Deficiency
- HP-II
- Pyrroline Carboxylate Dehydrogenase Deficiency
- Δ'-Pyrroline 5-Carboxylate Dehydrogenase Deficiency

Hyperprolinaemia Type 2 is a rare disorder characterized by a high level of an amino acid known as proline in the blood. This disorder is caused by a deficiency of an enzyme known as delta-pyrroline-5-carboxylate (P-5-C) dehydrogenase, the gene responsible (P5CDH) has been located on the short arm of chromosome 1.

There is no known treatment for this disorder, dietary treatment is not helpful however those affected with disorder seem to grow out of fevers and seizures. When the patient reaches adulthood, they appear to be symptom free.

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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