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RIBOSE -5-PHOSPHATE ISOMERASE DEFICIENCY [RPID]

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INTRODUCTION:-

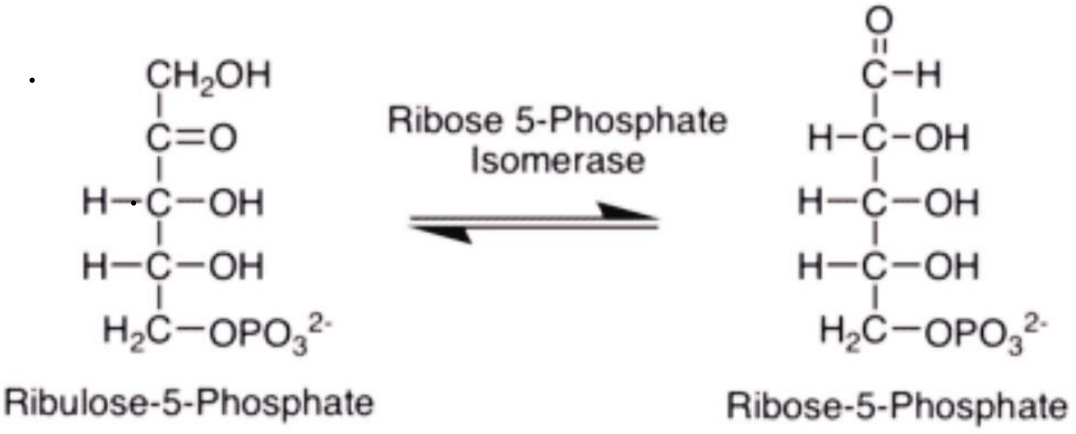
It is a rare human genetic disorder caused by inborn errors of mutations in the RPIA gene which codes to Ribose -5 phosphate Isomerase.

Ribose-5phosphate Isomerase [RPI] is an enzyme which involves in the carbohydrate degradation by pentose phosphate pathway.

RPI is a protein which exists in two forms i.e., RPIA and RPIB ,they can be found in prokaryotic and eukaryotic cells.

RPI helps in converting the Ribose-5-phosphate to Ribulose-5-phosphate.

RPI deficiency is mainly characterized by leukoencephalopathy i.e., deterioration of CNS white matter and an increased levels of ribitol and D-arabitol levels in the brain and body fluids .



ETIOLOGY:-

Mutations in the 'Hexose-Monophosphate-Shunt'[HMP SHUNT] pathway also called Ribose-5- phosphate Isomerase pathway.

DISCOVERY:

The first case has been reported in the year 1999 by Van der Knaap and team. They found that a 14 year old boy is suffering from delay in development , leukoencephalopathy, seizures, psychomotor retardation and

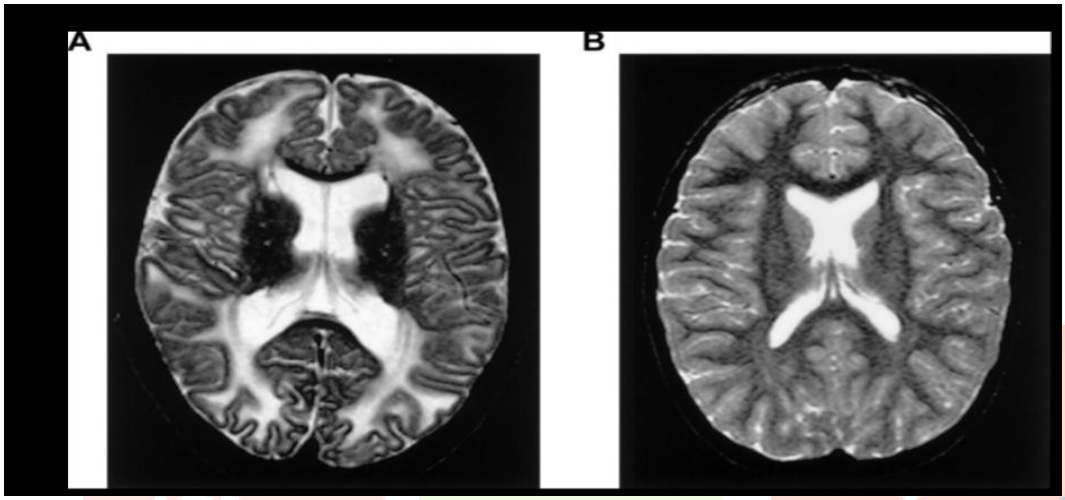
irregular polyol metabolism. The reason behind this was found to be frame shift mutation in the one of the allele.

The second case of RPI deficiency has been reported by Naik and fellow workers. They observed in an 18year old boy , who suffered with the same symptoms as that of the first case. The mechanism was found to be novel homozygous missense mutation and diffuse white matter abnormality.

The third case has been reported in 2018 by brooks and collegues. They found that in a new -born child with the symptom s of severe psychomotor regression and leukoencephalopathy.

Fourth case has been reported in 2019 by Kaur and collegues with the symptoms of progressive leukoencephalopathy and elevated polyols, aribitol and ribitol

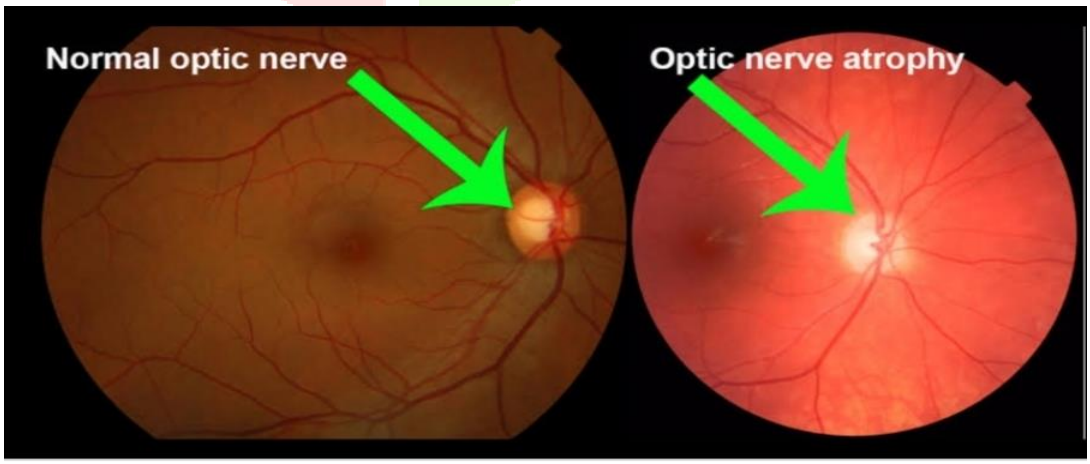
Therefore, with only 4 diagnosed cases , RPID is considered as the rarest disease.



The transverse t2 weighted image of the brain , showing extensive abnormalities of the cerebral hemispheric white matter in the patient,[A] as compared with that of a normal individual [B]

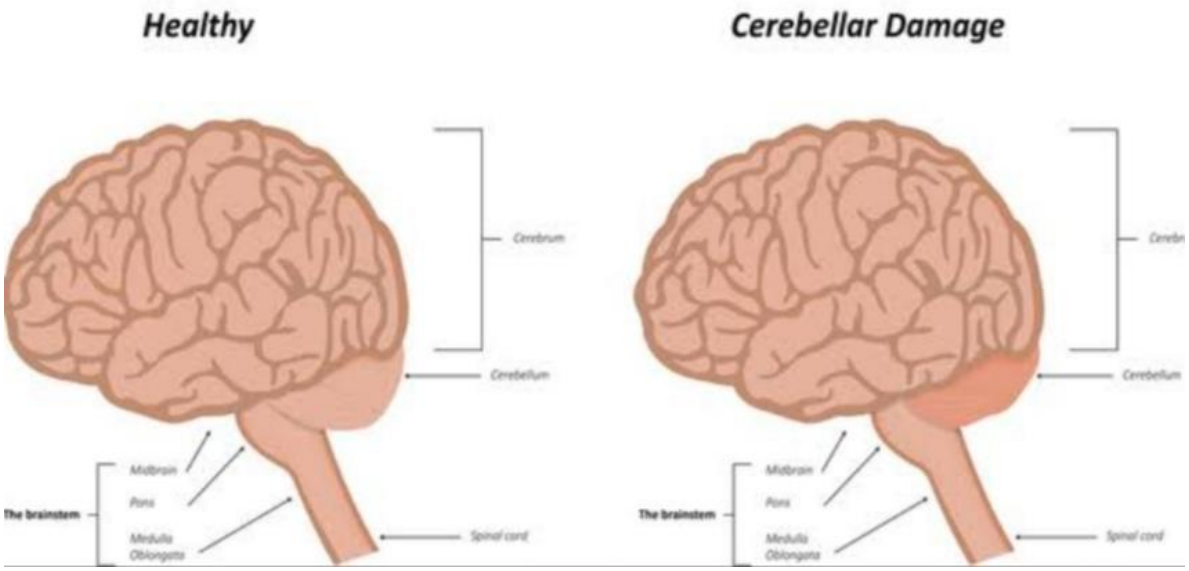
SYMPTOMS:-

- Optic atrophy:- death of nerve cells present in the eyes causing vision loss.

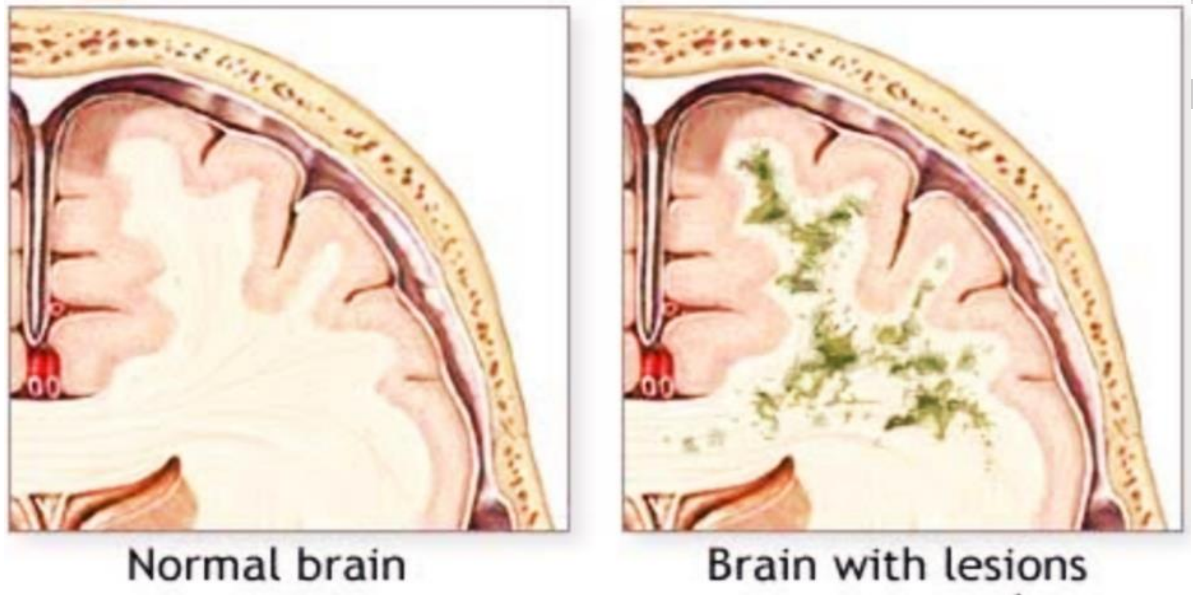


- Cerebellar ataxia:- uncoordinted muscular moment , because of cerebellum damage.

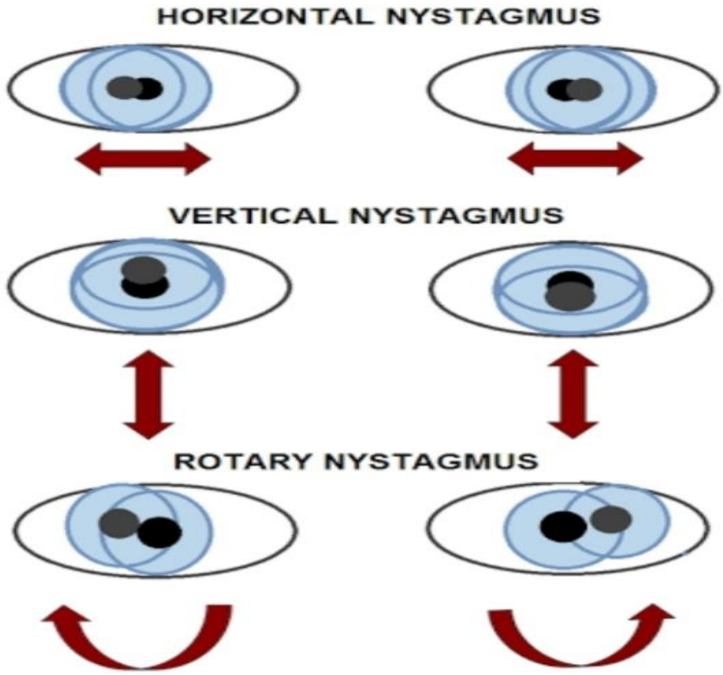
Cerebellar Ataxia



- Seizures:- disorder of the nerve cells present in the brain ,causes uncoordinated electrical disturbances.
- Spasticity:- alteration in the muscular performance which causes muscle stiffness and tightness.
- Leukoencephalopathy:-disease caused to the deterioration of the CNS white matter



- Nystagmus:- involuntary moment of the eyes



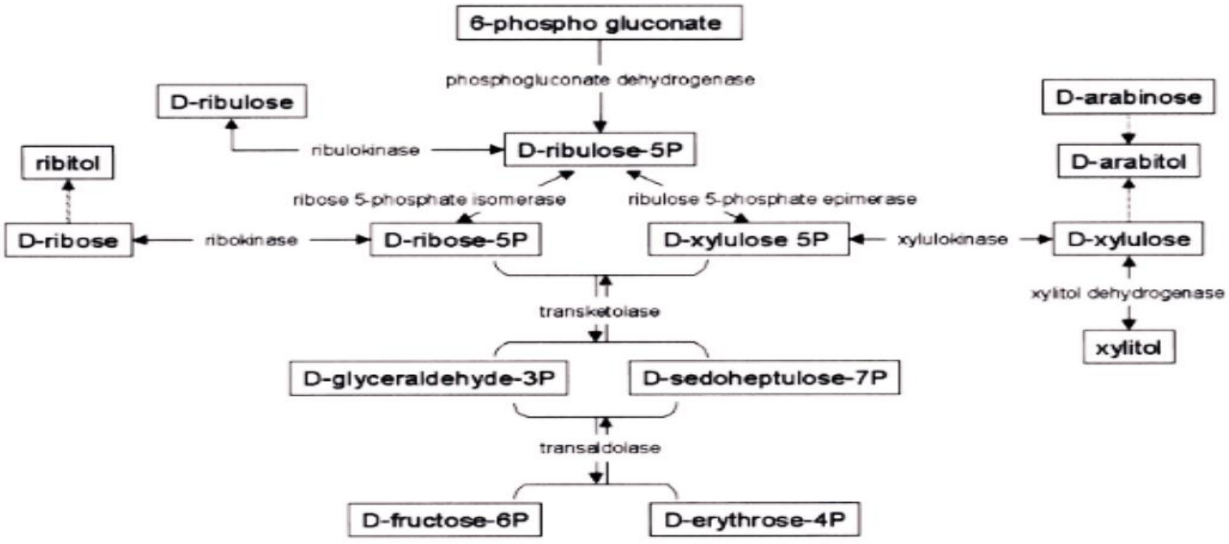
- Developmental delay:- disorder found in the developmental stages of the children characterized with the symptoms of delay in speech, cognition and daily life activities.
- Psychomotor retardation:- slow I physical activities, emotional activities and speech.

MECHANISM:-

RPI deficient patient found to have a rare allelic union, one of which alleles is non- functional and the other allele is partially active . so the partially active allele has the shortage of gene expression .Therefore, few patients will suffer from substantial amount of RPI activity.

Other reasons were found to prove that R5P is insufficient for RNA synthesis to occur.

It may also due to the secretion of D-arabitol and ribitol which gets secreted by the proton magnetic resonance spectroscopy of the braut the exact mechanism of RPID is still uknown.



DIAGNOSIS:-

- Based on the symptoms i.e., optic atrophy, cerebellar ataxia, seizures, spasticity, leukoencephalopathy, nystagmus, developmental delay, psychomotor retardation.
- Proton magnetic resonance spectroscopy of the brain
- Poluols analysis in the urine.

TREATMENT:-

There is no current specific treatment for RPI deficiency

But the symptoms of RPID can be managed by following ways:-

- OPTIC ATROPHY:-
  If it is diagnosed in the early stages , further damage of eyes cane be prevented.
- CEREBELLAR ATAXIA:-

  Episodic ataxia can be managed by giving ACETAZOLAMIDE drug and life style modifications can be suggested.

  Acquired ataxia can be managed by anti-micriobial and anti-viral medications

- SEIZURES:-
  Early diagnosis is suggested by the following tests such as EEG, CT scan, CAT scan, MRI scan.
  Treatment involves anti-epileptic drugs and weight reduction.
- SPASTICITY:-
  It can be decreased by performing stretching exercises daily and it also helps in improving motor flexibility.
- LEUKOENCEPHALOPATHY:-
  Avoiding drugs effecting immune system.
- NYSTAGMUS:-
  Regular eye check up is suggested to find the cause of action.

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