

Gene behind devastating vitamin disorder identified

An international team has identified the gene behind a rare but serious vitamin B12-related disorder and also picked up clues about how the vitamin acts within the body.

The rare genetic inability to process vitamin B12, usually diagnosed in infancy or childhood, may cause patients to suffer a host of debilitating problems like serious developmental delay, psychosis and anaemia.

Despite the variety of symptoms manifested by the disorder, this research shows that all of them are caused by mutations in different parts of the same gene, known as MMADHC.

'Most patients with B12 problems have difficulty absorbing the vitamin, or may be vegans who don't get it in their diet,' said David Rosenblatt of McGill University Health Centre, Montreal.

'However, this select group of patients becomes extremely sick because their bodies cannot transform the vitamin into its active forms.'

These findings have been published in the latest issue of the New England Journal of Medicine.

Vitamin B12 is an essential water-soluble vitamin found in dairy, eggs, meat, poultry, fish and shellfish, but not in plants. It is vital for the synthesis of red blood cells and the healthy maintenance of the nervous system. It helps control homocysteine levels, excess of which is associated with increased risk of heart disease, stroke and dementia.

The research relied heavily on expertise developed at McGill and University Children's Hospitals Basel as world referral centres for the diagnosis of B12-related genetic diseases, said Rosenblatt, a co-author of the study.

'This discovery offers earlier diagnosis and treatment options for this serious disease, and also helps explain the mechanism of how vitamin B12 works in everyone.'

Scientists at the University Children's Hospitals of Basel and Zurich in Switzerland, Brunel University in West London and McGill University and the McGill University Health Centre (MUHC) in Montreal were associated with the research.

(© IANS / India eNews)