Hallervorden Spatz Disease

An 18 years female presented with abnormal posturing of limbs with progressive gait impairment, slowing of voluntary movements, gradual diminution of vision and intelligence since the past one year. On clinical examination, there was generalised increase in tone in all four limbs and mild hyperreflexia. Fundoscopy revealed bilateral pigmentedary retinopathy. Laboratory investigations, which included urine metabolic screening, urine copper and serum ceruloplasmin were within normal limits. Her peripheral smear showed acanthocytosis. MRI done revealed the classic ‘eye of tiger’ appearance (Fig. 1). A diagnosis of Hallervorden Spatz was made based on clinical and MRI findings.

Hallervorden Spatz disease (HSD) falls in the category of Neurodegeneration with brain iron accumulation (NBIA), a group of progressive extrapyramidal disorders with radiologic evidence of focal iron accumulation in the brain. Inheritance pattern is autosomal recessive. Clinical features include early onset of progressive dystonia and intellectual impairment. Pigmentary retinopathy, choreoathetosis, pyramidal signs, optic atrophy, acanthocytosis are also frequently seen. Swaiman has described the clinical course as follows: (1) early onset childhood types; those with diagnosis before 10 years of life, either rapidly or slowly progressive (Type Ia and Ib), (2) Late onset types in which the diagnosis becomes apparent between 10 and 18 years (like our present patient) and (3) adult types. Characteristic pathological findings in HSD are neuroaxonal swelling and iron deposition in the globus pallidi of the lentiform nuclei.

MRI brain shows bilaterally symmetrical hyperintense changes (gliosis) with peripheral hypointensity (iron deposition) in the globus pallidi on T2WI [eye-of-the-tiger sign], a feature highly suggestive of this disorder. In 2001, Zhou et al linked HSD with a defect in the gene (PANK2) on the short arm of chromosome 20 (20p13) encoding the enzyme pantothenate kinase 2, involved in Coenzyme A synthesis. Hence this disorder was renamed Pantothenate Kinase Associated Neurodegeneration (PKAN). Oxidative stress due to accumulation of cysteine in the absence of pyrophosphopantothenate probably causes the damage. PKAN is the first identified disorder of pantothenate metabolism. To date the eye-of-the-tiger sign has been observed in all patients with PANK2 mutations. It is a fairly specific sign and has not been seen in other non-PKAN NBIA as neuroferritinopathy and aceruloplasminemia. This sign can be used to identify patients for PANK2 genetic testing and has accurately identified presymptomatic siblings of affected children.

Oral medications that seem to provide the most relief include baclofen and trihexyphenidyl. L-dopa/carbidopa and bromocriptine can be of some help. Therapies that have been tried to manage dystonia in some individuals with varying success include intramuscular botulinum toxin and intratechal baclofen. Stereotactic pallidotomy and bilateral thalamotomy occasionally have been tried for patients with severe dystonia, resulting in partial relief of symptoms.

SB Sabat*, MP Deshmukh**

*Lecturer, Department of Radiology and Imaging, Grant Medical College and Sir JJ Group of Hospitals, Bculla, Mumbai - 400 008; **Consultant Radiologist, Lilavati Hospital and Research Centre, Bandra, Mumbai - 400 050.

Received : 4.4.2006; Revised : 25.5.2006; Accepted : 7.7.2006

REFERENCES