ORIGINAL RESEARCH PAPER

Medicine

Volume - 7 | Issue - 4 | April-2017 | ISSN - 2249-555X | IF : 4.894 | IC Value : 79.96

CINCA Syndrome (Chronic Infantile Neurological Cutaneous Articular Syndrome)

KEYWORDS

Aparna Vishal More

Assistant Professor at Rajiv Gandhi Medical College, Kalwa.

ABSTRACT CINCA syndrome is a genetic disorder characterized by early onset recurrent episodes of fever, headache, and rash, progressive articular & neurological involvement. We report a case of 16 years old girl with CINCA syndrome with history of recurrent headaches & fever. She had a de novo F309S mutation in exon 3 of CIAS1 gene on chromosome 1. This article is of interest to physician as it is a rare unknown disease.

Case report

A 16 years old girl born of non-consanguineous marriage was admitted in wards for complains of recurrent headache, fever with skin rash & joint deformity with restriction of movement. She had sparse clinical records with multiple previous admissions with prescription of analgesics, local ointments, antibiotics & intermittent oral steroids.

On examination the patient was conscious cooperative, obeying commands. Patient was febrile with pulse of - 120/min, Blood pressure – 120/80mmHg. Patient had neck stiffness so CSF examination was done. CSF report showed increased proteins with increased lymphocytes suggestive of tuberculous meningitis. Patient was started on Anti tuberculous treatment. Skin reference was taken suggestive of erythematous rash all over body. Funduscopic showed retinal vasculitis. Other systemic examination was unremarkable.

Complete blood count showed Hemoglobin of 7g/dl, TLC 12000cells/cu.mm, Platelet count normal, ESR – 110. Urine report was normal, Skin biopsy done showed atrophic epidermis & dermal inflammation with perivascular neutrophilic infiltration with no granuloma. USG of abdomen suggestive of hepatosplenomegaly. X-ray chest was normal. X-ray of lower limb showed enlarged deformed femoral bone, patella & tibia with valgus deformity, the patella had heterogeneous ossification.

Due to clinical profile & radiological findings compatible with diagnosis patient was investigated further. Sequencing of C1AS1 gene amplified from genomic DNA isolated from peripheral blood leucocytes revealed F309S mutation in exon 3 on chromosome 1[5].

Since patient had CSF picture suggestive of tuberculous meningitis, so patient was started on anti-tuberculous with steroids.

Discussion

The disease often stays soon after birth as in our patients & lasts of life long [1]. Recurrent flares consist of fever, nonpruritic urticarial rash, lymphadenopathy and hepatosplenomegaly characterize the course. Articular manifestation vary from arthralgia, transient swelling without effusions to severe deforming arthropathy. Premature patellar ossification with patellar overgrowth is frequent & was important clue in our patient [6]. Saddle back neck, frontal bossing, short hands & feets with fingers clubbing are the peculiar morphologic characteristics seen. Ocular manifestations can be retinal vasculitis [2,4]. Neurological involvement can evolve over childhood or may have delayed onset [3]. Non-steroidal anti-inflammatory drugs & corticosteroids offer temporary relief for pain, fever, joint mobility. Various drugs such as azathioprine, colchine, cyclosporine, etanercept, infliximab, intravenous immunoglobulin, methotrexate, penicillamine, Salazopryin, thalidomide have inconsistent results. At present Anakarin an IL- 1, receptor antagonists the most promising medications for control of symptoms. Our patient was started on steroids plus anti tuberculous line of treatment.

References:

1. Report of a child with neonatal-onset multisystem inflammatory disease and review of the literature Zhonghu Er Ke Za Zhi. 2014 Dec; 52(12):932-6.
2. Cryopyrin-associated periodic syndromes and the eye Oberg TJ, Vitale AT, Hoffman RO, Bohnsack JF, Warner JE. Ocul Immunology Inflamm. 2013 Aug; 21(4):306-9.
3. Neurologic manifestations of the cryopyrin-associated periodic syndrome. Neurology. 2010 Apr 20; 74(16):1267-70. Doi: 10.1212/WNL.0b013e3181d9ed69.
4. CINCA syndrome: a rare cause of papilledema. The case of homozygous twins]. J Fr Ophtalmol. 2010 Jan; 33(1):36-9. Doi: 10.1016/j.jfo.2009.11.003. Epub 2009 Dec 10.
5. Phenotype-genotype analysis of cryopyrin-associated periodic syndromes (CAPS): description of a rare non-exon 3 and a novel CIAS1 missense mutation. J Clin Immunol. 2008 Mar; 28(2):134-8. Epub 2007 Dec 15.
6. CAPS: cryopyrin-associated periodic syndrome Nihon Rinsho Meneki Gakkai Kaishi. 2011; 34(5):369-77.