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Rare Case of Roger Syndrome

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INTRODUCTION

Roger syndrome (Thiamin Responsive Megaloblastic Anemia Syndrome), is characterized by megaloblastic anemia, deafness and diabetes mellitus. It is an autosomal recessive disorder due to mutation in SLC 19A2 gene that leads to defective thiamine transport protein. So, thiamine is not absorbed and effectively utilized by body from diet.[1]

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CASE REPORT

We report 9.5 years old male child product of consanguineous marriage, fully vaccinated according to EPI schedule presented in Faisalabad Medical University / Allied Hospital, Faisalabad with history of:

Recurrent blood transfusion for 7 years. Uncontrolled diabetes for 4 years.

Patient actual history dates back to 2 years of age when mother noticed pallor, gradual in onset with no H/O bleeding from any site, jaundice, bone pains, chronic diarrhea, any urinary complaints, joints pain, rashes, abdominal pain, H/O pica or passage of worms in stool. Child got first blood transfusion after 2 months of developing pallor as advised by local doctor without proper investigations and since then having recurrent blood transfusion initially every 3–4 months and later reduced to every 4 weekly for last one year. Pre-transfusion Hb dropped <7gm/dl before each transfusion.

Child started having polyurea and polydipsia at age of 5 years and was diagnosed as type I DM and advised insulin 70/30 in 2 divided doses by some general practitioner. Patient was compliant with therapy but had poor control of blood sugar.

H/O delayed speech and only says 1 – 2 words up till now and parents noticed at 3 years of age that child has no response to sounds.

Child was diagnosed antenatally as having hydrocephalus. Ventriculoperitoneal shunt was done at 20 days of life due to obstructive hydrocephalus.

H/O hydrocephalus in elder sibling, who died at age of 9 months. Two other siblings alive and healthy socio-economic status is poor, father is a factory worker.

Patient developed neck holding at 8 months of age, sitting at 7-8 months, standing 1.5 year and walking about 2 years

Patient was on breast feeding for up to 1 year, weaning started at 6 month of age and now taking average diet.

EXAMINATION

9.5 years with obvious pallor having pulse 110/m, RR: 30/m, temp 98^{o}F, B.P: 100/70 mmHg, Hight: 117 cm & weight: 23 kg, (below 5th centile) OFC: 52 cm.

No jaundice, clubbing, rash, edema, bruises, petechiae, lymphadenopathy, skeletal, deformity, hyperpigmentation.

Systemic examination is unremarkable.

INVESTIGATIONS:

Patient was previously investigated by local doctors and had repeated CBCs with him that showed low Hb and RBC count and high MCV in most of the reports. Platelets and WBC counts were normal.

Serum IgA and antitissue transglutaminase IgA level were also normal. S. Ferritin was 39.8 ng/ml (20-300ng/ml). Serum folic acid and B12 level were also within normal range.

Stool for occult blood was negative.

ECG and echocardiography were normal.

Bone marrow biopsy showed good cellularity and hematopoiesis with adequate erythropoiesis, micro normoblasts and megaloblasts. Myelopoiesis and megakaryopoiesis was also adequate.

Audiometry results obtained from Free Field Testing in controlled environment reveal bilateral moderate degree hearing loss. Sound awareness thresholds are obtained at 45-70 dbHl.

On the basis of clinical picture and investigations, patient was diagnosed as Thiamine Responsive Megaloblastic Anemia (Roger syndrome). Sample was sent for mutation analysis to National Institute for Biotechnology and Genetic Engineering (NIBGE) and results are awaited.

Patient was started treatment in the form of Inj. Neurobion containing 100mg thiamine daily intramuscular. (as no pure thiamine preparation is available in Pakistan).

Patient was also started Inj. lispro and long acting glargine regimen.

Now for last 8 months, patient has no need of blood transfusion and his blood sugar level are also controlled. His latest Hb is maintained on 12g/dl. Patient is now shifted to Tab. Neurobion.

DISCUSSION

TRMA is described by Roger in 1961.[2] Thiamine in a dose of 25–75 mg/d is required for treatment of megaloblastic anemia in

pediatric population. On stopping of thiamine, anemia was again reported. High doses of thiamine can delay presentation of diabetes mellitus.[3] This response of treatment was also noticed in our patient as need of blood transfusion was no more required on start of thiamine.

TRMA is an autosomal recessive disorder caused by deficiency of thiamine transport protein.[4] Thiamine is water soluble vitamin which is used for tissue growth and development. Deafness that is irreversible to treatment, megaloblastic anemia and diabetes mellitus are cardinal features of roger syndrome.[5] Arrhythmias and structural abnormalities of heart are also associated with it.[6] On bone marrow biopsy, there will be megaloblastic anemia with occasional ringed sideroblasts. A mutation in SLC19A2 gene have been reported in patient with TRAMA, it is required for thiamine affinity transporter.[7] There should be annual follow up to see efficacy of oral thiamine and disease progress in form of some blood test which includes CBC, Retic count, Hb1AC, hearing, visual and cardiac assessment.[8] Prenatal diagnoses of Roger Syndrome is also available for families.[9]

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