



A case report on Menkes Kinky Hair disease with bronchopneumonia, generalized tonic clonic seizures, global developmental delay and failure to thrive.

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Abstract:

A 13-months-old male infant with complaints of fever, cough and cold since 7 days and was having 8-10 episode of generalized tonic clonic seizures which lasted for 5-10 second each. The history of infant has generalized tonic clonic seizures from last 8 months. The infant had delayed developmental history and weighed 6 kg on the day of admission. The infant had fair complexion with fuzzy, brittle, and sparse hairs over head with very low pitch voice and was not able to open eyes. The patient showed low levels of copper and higher CRP(C-reactive protein) and ESR values. The infant was diagnosed with bronchopneumonia, GDD, FTT, GTCS and Menkes kinky hair disease. The infant was given symptomatic therapy.

Introduction:

Menkes disease is an X-linked lethal multi system disorder, which occurs due to mutation of 'p-ATPase7' gene which causes disturbances of copper distribution in different tissues. The estimated prevalence is 1 in 1,00,000 to 1 in 2,50,000 [1]. The patient suffers from defect of copper containing enzymes resulting in multi systemic disturbances. Nervous system problems consist of gross mental retardation, convulsions, asymptomatic subdural effusion, cortical atrophy, gross trunkal hypotonia and progressive neurological deterioration,

Vascular problems with weak collagen tissues causes fragility, connective tissue abnormality gives rise to characteristics steel, fuzzy, wooly, sparse hair which are easy to pluck out. [2]

The bones are osteopenic. There are lot of chances of recurrent infections and so the infant fails to thrive and malnutrition is a common finding [3]. The infant usually dies within 3–4 years of age. In 1962, Menkes first described the syndrome and Drank et.al. noted the association with copper metabolism [4]. The affected gene was cloned in 1993.

Case Report:

A 13-months-old infant was admitted to hospital with chief complaints of high grade fever, intermittent, relieved on medication since past 7 days, complaints of cough and cold since past 7 days and 8-10 episodes of convulsion which lasted for 5-10 seconds each with up rolling eyeballs. The infant had a history of convulsion since past 8 months and was on OPD based medication. The infant was not able to hold neck as well as sit, even at age of 13 months; this shows signs of developmental delay in the patient. The infant was immunized up to MR but no documents were available. The infant was on breast feeding till the age of 10 months; since past 1 month, infant was started with complete feeding of rice, milk, biscuit, and khichadi.

General Examination:

On examination, infant was conscious but irritated, fair complexion with chubby cheeks and curly light brown hair. Anthropometric measurement revealed weight: 6 kg, height: 79 cm, head circumference: 42 cm; there was mild pallor; skull, spines and bones were normal. Pulse: 120bpm, temperature: 101^o F, respiratory rate: 50 p/m, SpO₂: 98%, RBS: 91 mg/dl. Central nervous system examination revealed repeated generalized tonic clonic seizures, the trunk was hypotonic. Examination of Respiratory system showed features of bronchopneumonia. Other systems appeared normal.

Laboratory Investigation:

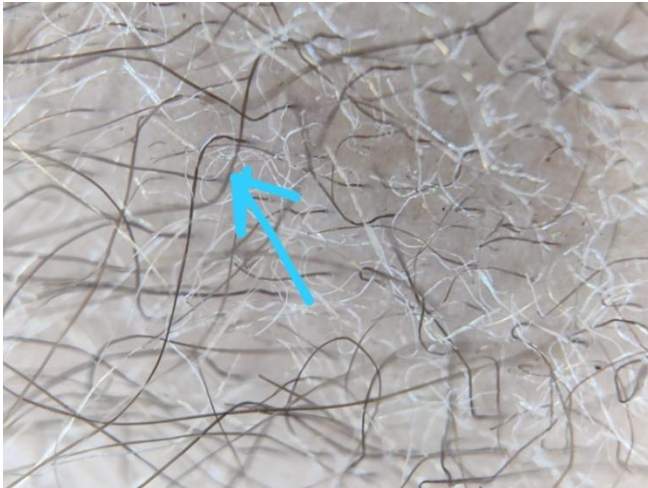
On investigation the infant was found suffering from mild anemia (Hb: 9.50 gm/dl), lower red blood cell count (4.13/cmm), high WBC count (13000 /cmm), high lymphocytes (54%), lower PCV (29.90%), lower MCV (72 fl), lower MCHC (23.10 pg).

The infant's C-Reactive Protein was 12 mcg/ml which was positive and Erythrocyte Sedimentation Rate was 60 mm.

The Serum Copper level was 29.59 mcg/dl and Ceruloplasmin Serum was 19.74 mg/dL while on 10th of treatment infant SGOT was 177 IU/L

Infants MRI- BRAIN showed Benign enlargement of subarachnoid spaces along bilateral fronto-parietal lobes. Arteries of circle of Willis, appears mildly elongated and tortuous. Mild prominence of vessels in the basal ganglia on both sides. Microscopic view of hair

reveled pili torti, trichoscopy which reveled hypopigmentated white brown hair with short length.



The therapeutic plan of medication:-

Drug	Dose/route /frequency
Inj. AUGMENTIN	180 mg/ IV/ 8 hrly
Inj. AMIKACIN	30 mg /IV/8hrly
Inj. PCM	60 mg/ IV/ SOS
Inj. ½ DNS + 5CC KCl	25 cc/hr / IV
Syp. LEVETIRACETAM	2 ml / PO / HS
Syp. ZINC	2.5 ml /PO /OD
Syp. Ca ⁺⁺ + D3	2.5 ml /PO/BD
Syp. NERVITAS GOLD	3.5 ml /PO / BD
Inj. ELDERVIT	0.5 ml /IM /OD
Syp. POTCHLOR	5 ml /PO / TDS
Tab FA	½ Tab / PO/OD
T. BIOTIN	5 mg / PO / BD
Inj. MgSO ₄	0.5 ml

Discussion:

Menkes disease is an X – linked disorder, which results in profound systemic copper deficiency. The Menkes disease gene is (ATP7A) which encodes an enzyme p-type ATPase which is required for systemic absorption, distribution and metabolism of copper in tissues. There is no racial bias. The disease usually manifests at the age of 2–3 month and individual usually die at 3–4 years of age due to pneumonia. Our patient presented at the age of 13 months.

Menkes disease causes developmental delay, seizures, hypotonia which is more in trunkal muscle and difficulty in feeding. All these are marked in our patient. The characteristics facies, fair complexion, chubby cheeks, sparse twisted fuzzy depigmented hair are all present in our case. The investigation finding of low serum copper, ceruloplasmin, hair structures are characteristics of Menkes disease[8]. MRI studies are supportive of the disease. The growth failure in our case is also a common finding of this syndrome.

The higher levels of copper, the microscopic view of hairs and the malnutrition of the infant and the investigation data diagnosed the infant as Menkes Kinky Hair Syndrome with bronchopneumonia, GDD, FTT and GTCS.

Discharge Discussion:

On 14th day infant was planned to discharge due to improving condition of infant. The discharge medication was given as followed:

Syp PCM 4 ml / PO / SOS

Syp ca⁺ + D3 2.5 ml / PO / BD

Syp. LEVETIRACETAM 1 ml / PO / BD

Syp ZINC 2.5 ml / PO / OD

Medication was given for 10 days and advised on follow up after 10 days and was planned for prenatal genetic counseling on follow up.

Abbreviation:

GDD Global Developmental Delay, FTT Failure To Thrive, GTCS Generalised Tonic Clonic Seizures, WBC White Blood Cells, PCV Packed Cell Volume, MCV Mean Corpuscular Volume, MCHC Mean Corpuscular Haemoglobin Concentration, Hb Haemoglobin.

Conclusion:

Menkes disease is a rare condition so one needs to be aware about it and it's clinical presentations. Diagnosing menkes early and managing it's complications is very important. Clinical suspicion of possibilities may lead to accurate diagnosis.

Compliance with ethical standards:

Acknowledgments

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Disclosure of conflict of interest All authors declare that they have no conflict of interest.

Statement of informed consent Informed consent was obtained from the individual participant included in the study.

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