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A Case of Megaloblastic Anemia With Suspected Gilbert Syndrome

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Clinical history:

A 27 year old male patient came with complaints of generalized weakness since 15 days associated with nausea and reduced appetite.

He also complained of fever, low grade, intermittent in nature with no diurnal variation.

No history of loose stools.

No history of joint pains or rashes.

k/c/o GILBERT SYNDROME- diagnosed two years ago (details not available)

Examination:

GENERAL PHYSICAL EXAMINATION:

Afebrile.

Pallor and icterus present.

No generalised lymphadenopathy noted.

PER ABDOMEN:

Inspection:

Shape of abdomen: normal

Umbilicus central.

No distention visible

Palpation :

No local rise of temperature or tenderness noted.

Spleen is palpable 3cm below the left costal margin.

Percussion: Traube space is obliterated-dullness noted.

No Signs of free fluid.

Auscultation. Normal bowel sounds heard.

CVS: S1, S2 heard. No murmurs.

RS: B/L NVBS heard. No added sounds.

CNS: Within normal limits

Investigations:

BLOOD INVESTIGATIONS:

Hb-5.5gm%

TLC-3670 cells/cumm

N-47.9

L-43.9

RBC count-1.47million/cumm

MCV- 109fl

MCH -37.3 pg

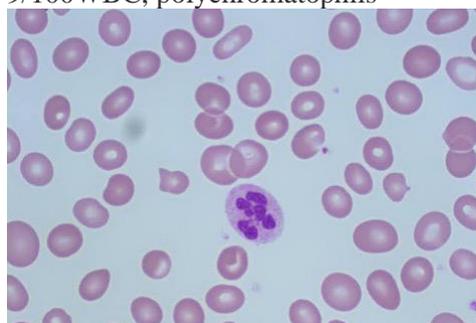
MCHC -34.3 g/dl

Platelet count-44,000/cumm
Direct coombs test-negative
Retic count-6%
Vit B12-1254pg/ml (190-950pg/ml)

Folic acid-1.2ng/ml(2-20ng/ml)
LDH-1535U/L (125-220U/L)

PBS picture:

Pancytopenia with anisopoikilocytosis, fragmented RBCs, basophilic stippling, tear drops cells noted. 9/100WBC, polychromatophils



Blood picture of megaloblastic anemia-macroovalocytes with hypersegmented neutrophils.

Ref: <http://doi.org/10.7939/R36970D1B>

LIVER FUNCTION TEST:

Total bilirubin -3.53mg/dl
Direct bilirubin-0.46mg/dl

Diagnosis:

- Pancytopenia due to Megaloblastic anemia secondary to nutritional deficiency
- Gilbert Syndrome ??

Treatment:

- Patient was treated with 1500 micrograms of Vitamin B12 injection and 5 mg of folic acid tablets.
- Two pint PRBC transfusion was given after which his Hb improved from 5.5gm% to 9.3gm%.
- Patient was discharged with the advice to continue B12 and Folic acid.

Discussion:

Megaloblastic anemia results from the deficiency of vitamin B12, or folic acid or from disturbances in folic acid metabolism. Folate is an important substrate of and vitamin B12 an important co factor for the generation of essential amino acid methionine from homocysteine. Deficiency of vitamin B12 or folate will therefore produce high plasma levels of homocysteine and impair DNA synthesis. The end result is cells with arrested nuclear maturation but normal cytoplasmic development-so called nucleocytoplasmic asynchrony.¹

High proliferation rate of bone marrow results in striking changes in hematopoietic system in megaloblastic anemia. Cells become arrested in development and die within the marrow, this ineffective erythropoiesis results in an expanded hypercellular bone marrow.¹

Macrocytic anemia with increased mean corpuscular volume (MCV), defined as more than 100 fL, is the hallmark of megaloblastic anemia, but leukopenia and thrombocytopenia are also frequently present.² Megaloblastic anemia is the commonest cause for pancytopenia

Since Megaloblasts are prone for hemolysis, LDH is increased. This could be the cause for indirect hyperbilirubinaemia. There is every possibility that the patient could have been wrongly diagnosed to be having Gilbert Syndrome.

Gilbert syndrome is an autosomal dominant trait caused by mutation in the promoter region of UDP-glucuronyl transferase enzyme, which leads to reduced enzyme expression. This results in decreased conjugation of bilirubin, which accumulates as unconjugated bilirubin in the blood.¹

Typical presentation is with isolated elevation of bilirubin, typically, although not exclusively, in the setting of physical stress or illness.¹

The only significant laboratory abnormality in patients with Gilbert syndrome is increased unconjugated bilirubin levels, and they are usually below 3 mg/dL with less than 20% of the bilirubin levels being conjugated. When associated with other pathological conditions which increase hemolysis, the level can be higher, but even then it is usually below 6 mg/dL.³

References:

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3. Viveksandeep T, Savio J. Gilbert syndrome. [Internet]. 2020 [cited 7 March 2020];. Available from: <https://www.ncbi.nlm.nih.gov/>