



Iron-Refractory Iron Deficiency Anemia (IRIDA) a Hypochromic Microcytic Anaemia with Iron Deficiency

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Introduction

Iron Deficiency Anemia (IDA) is a frequent entity that generates 50% of the anemia in the world. Even if the etiologies of the IDA are often due to nutritional deficiency, bleeding or gastrointestinal malabsorption; genetically inherited iron deficiency anemia should also be kept in mind if there is an unknown cause of iron deficiency anemia.

Iron-Refractory Iron Deficiency Anemia (IRIDA) is a hypochromic microcytic anemia, an autosomal recessive disorder and shows iron deficiency anemia that is refractory to oral iron therapy but partially refractory to parenteral iron therapy. This hypochromic microcytic pattern connected with the low transferrin saturation, normal or high ferritin and abnormally high hepcidin levels. The underlying reason for the disease is the mutations of the *TMPRSS6* gene. Normally *TMPRSS6* gene encodes a trans membrane serine protease matriptase-2 that has negative effect on the production of hepcidin iron regulatory protein. Hepcidin is the equalizer of the ferroportin, the main iron supplier. In *TMPRSS6* gene mutation, matriptase-2 protein cannot be produced. Thus, hepcidin levels increase and inhibit ferroportin. Even though the iron storage exists, the iron that cannot enter to the systemic circulation causes iron deficiency anemia and this anemia is refractory to oral iron therapy.

Here we present a patient diagnosed as iron deficiency anemia refractory to iron and confirmed with the *TMPRSS6* gene analysis. The patient had urinary tract infections two times in her early childhood but did not repeat later on. Her urine examination was normal and culture was negative. Patient's grandfather has urolithiasis and her aunt has gravel, therefore the patient is followed in this respect. Plenty of liquid and nutrition advice were given to the patient and 3 months later USG repetition is planned.

Erythrocytes are cells responsible mainly for gas exchange process, so it is necessary a high degree of deformability in order to facilitate their passage through reduced caliber vessels and thus oxygenate tissues. This deformability is conferred by several factors such as a high area-volume ratio (conferred by the biconcave disc shape) and the degree of fluidity of the plasma membrane. Thus, deformability is one of the most important features of a mature erythrocyte, with several factors affecting it. PI3K/Akt signaling pathway in erythrocytes along with nitric oxide synthetase showed to improve

deformability. Additionally, rho kinase plays an important role in erythrocyte deformability when a specific inhibitor, Y-27632, increased erythrocyte deformability by inducing ATP release from these cells.

AMPK Activity is Suppressed by TSH Treatment

Blood was collected in 4 mL EDTA tubes. These tubes were centrifuged for 10 minutes at approximately 3,000 rpm. After this, the plasma and buffy coat was removed and discarded. The RBC was washed with PBS (phosphate buffered saline) three times (tubes were homogenized by inversion and centrifuged in the same parameters as above). Wide range of NaCl concentrations (0.1%-0.9%) was added, followed by TSH in four concentrations (1 mIU/L, 2 mIU/L, 3 mIU/L and 5 mIU/L), and RBC, resulting in an experiment with a final volume of 300 μ l and 1% hematocrit. The tubes were placed in the water bath, 37°C, and all experiments was incubated for one hour with hormone whether combined or not with inhibitors or activators, centrifuged for 10 minutes, 10,000 rpm and the supernatant were carefully removed and pipetted (200 μ l) into the 96-well plate. The reading was made in an Elisa reader (Tecan GENios®) at 540 nm absorbance.

TSH is classically associated to CAMP/PKA and PLC/PKC signaling pathway and others studies points to alternative pathways like activation of ERK 1/2 and NF κ B. In an attempt to elucidate possible signaling pathways that can be triggered by TSH in this specific system, we added AICAR, an AMPK activator, to assess whether the late is involved in TSH- mediated effects. We notice that AICAR blunted the protection in all concentrations studied, indicating that AMPK activity is suppressed by TSH treatment. This is in accordance with literature when Shudong et al. reported that AICAR blocks the TSH- induced expression of specific genes in liver. Besides, Thali and co-workers reported the presence of AMPK in red blood cell lysates and AMPK-deficient mice exhibited an increased hemolysis resistance around 150 msmol/L (correspondent to 0.4% NaCl) when compared to wild-type control model.

Excisional Biopsy with Careful Pathologic Review

Castleman's disease is a great mimicker of multiple illnesses due to its various manifestations. In this case, the plasma cells on the core biopsy misled providers to a diagnosis of myeloma. An initial excisional biopsy with careful pathologic review is required to properly capture the nodal architecture. This delay led to the aggressive widespread dissemination of the disease requiring a 5-drug regimen which is an uncommon approach in this disease. The disseminated KS, presenting as rapid painful rashes instead of singular purple skin lesions, also deceived the medical team. This case illuminates the diagnostic complexity of lymphadenopathy workup in HIV+ patients and highlights the importance of pursuing ample tissue in order to make an accurate and timely diagnosis.

Transfusion protocols today are centered on hemoglobin of 7 g/dL, although case reports involving Jehovah's witnesses and select trials have provided insight into the outcomes of life-threatening anemia. Carson et al. described a 33% mortality for patients with a preoperative hemoglobin of less than 6 g/dL, and additional studies have shown a 44% mortality without transfusion for a hemoglobin less than 5 g/dL. Within non-surgical patients, hemoglobin of 5 g/dL has

been showed to be tolerated in healthy euvolemic individuals, although this threshold is not well understood in the critically ill, elderly, or patients with decreased cardiovascular reserve.