NOVEL HEMOGLOBIN RATCHATHEWI: FIRST DISCOVERY AS A CAUSE OF LOW OXYGEN SATURATION FROM PULSEOXIMETRY

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Objective: To report the novel 'Hemoglobin Ratchathewi' case series

Methods: We performed the case series report from the data of Ramathibodi Poison Center (RPC) Toxic Exposure Surveillance System and Ramathibodi hospital.

Results: The consulting cases to RPC, with 'the oxygen saturation gap' or 'the difference between the low measured oxygen saturation from pulse oximetry and the normal calculated oxygen saturation from the arterial blood gas', were analyzed. The conditions of methemoglobinemia or sulfhemoglobinemia were excluded. All cases were referred to work up more at an outpatient clinic, Ramathibodi hospital. Hemoglobin typing with thalassemia DNA analysis and DNA sequence analysis were investigated. There were 2 families of rare hereditary hemoglobin anomalies discovered as the cause of "saturation gap". These included 1 hemoglobin Cheverly family and 1 hemoglobin La Desirade/Louisville family. Interestingly, we found 4 index cases and their other 6 family members (totally 10 cases) who had the novel hemoglobin. Their hemoglobin DNA sequence analysis shows the nucleotide variant as nucleotide substitution at codon 88 of alpha1 globin gene, 'histidine to glutamine'. To our knowledge, this nucleotide variant has never been reported in the literatures before. We proposed that this hemoglobin is the first and new report and it was named as 'Hemoglobin Ratchathewi' (the name of the district of Ramathibodi hospital's location). One case in our series was detected both nucleotide variants; 'Hemoglobin Ratchathewi, and hemoglobin Westmead'. All cases were healthy and did not look obvious cyanosis. All had the low measured oxygen saturation from pulse oximetry with different values of oxygen saturation (70-85%). No evidences of methemoglobinemia and hemolysis were found in these cases. Every case received the genetic counselling.

Conclusions: Hemoglobin Ratchathewi is the novel hemoglobin which is the first report in the cases with "the oxygen saturation gap". The physiology and clinical course of this new hemoglobin should be studied further. This hemoglobin identification is crucial for genetic counselling, moreover, avoiding unnecessary investigation and treatment.