Figure 1: The patient's peripheral blood smear. The black arrow shows a Knizocyte. The blue arrow shows a normal red blood cell

DISCUSSION

According to literature, only few cases of SAO had been reported in Sri Lanka. Most of the SAO patients are asymptomatic, but has been reported to be associated with signs of mild haemolysis such as intermittent jaundice, gallstones [4], and babies have been reported with neonatal hyperbilirubinemia [6].

Individuals who are homozygous for the SAO are not clearly identified and thought to be lethal. Anyhow, the study conducted by Liu S.C. observed, that none of the children were homozygous in families where both parents were heterozygous for SAO27bp deletion [3]. Those families had high rates of miscarriages [3]. However, Picard Veronique, recently reported a case of homozygous SAO where extremely severe dyserythropoietic anaemia associated with distal renal tubular acidosis. It was in a child born to asymptomatic Comorian parents. On 22 weeks of gestation, this male foetus presented with hydrops and severe anaemia (Hb of 2.9g/dL) and had to be treated with in-utero transfusions. Even after birth he needed monthly blood transfusions to keep the haemoglobin level between 7 and 10g/dL [7].

Diagnosis can be done by molecular genotyping where genetic analysis conducted to identify the mutation in SLC4A1 gene. Light microscopic diagnosis is also a possibility. Red blood cell morphology in the peripheral blood smear shows the presence of ≥25% ovalocytes and some stomatocytes[8]. The red blood cells of SAO are often described as being stomatocytic elliptocytes with a slit-like area of central pallor. In a small proportion of these stomatocytes, 2 or less frequently 3 pale regions separated by a well haemoglobinated ridge are apparent, giving the appearance of double stomatocytes, known as "Knizocytes" [9]. This feature is not pathognomonic for SAO but this morphology is the key to accurate diagnosis of SAO by a stained peripheral blood smear [9]. In a study by O'Donnell, red cells with 2 or more linear or irregular shaped pale regions (knizocytes) were also the most consistent feature of SAO in microscopy [10].

Most of cases of SAO are asymptomatic and treatment is not necessary. For the once who presents with uncompensated haemolytic anaemia, splenectomy can be considered, as some patients undergo remission with splenectomy [11].

Prognosis in heterozygous form is good and not life threatening. However, prognosis is uncertain in homozygous individuals [7].

Author declaration

Acknowledgements

We thank the patient for her interest and support, the staff of the medical unit for their valuable help in the management of this patient and all the members involved in hematology and chemical pathology laboratories in District General Hospital, Negombo for the technical skills.

Author Contributions

Nilshan Fernando was involved in the initial conception of the study and collected and interpreted clinical and biological data. Champa Jayamanna treated the patient. Kavindya Fernando was involved in data analysis, planning of the study and drafted the manuscript. All three authors revised the final manuscript.

Funding sources

No sources of funding were involved in this paper

Availability pf data and materials

Pictures of the blood smears used in the study are freely available from the corresponding author.

Ethics approval and consent to participate

Informed written consent to publish the details of the patient was taken from the patient herself.

Competing interests

We declare no competing financial interests

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