Incidence of Thalassaemia and Haemoglobinopathies with Normal Red Cell Indices – A Case for Universal Antenatal Thalassaemia Screening

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AIM

Thalassaemias and haemoglobinopathies are autosomal recessive disorders. In the absence of universal screening, asymptomatic carriers are only screened when presenting with abnormal red blood cell (RBC) indices. However, it is known that some carriers can present with completely normal RBC indices and not be identified for screening as a result. We recently implemented universal antenatal thalassaemia screening in KK Hospital. This poster aims to describe the incidence and spectrum of haemoglobinopathies and thalassaemias presenting with normal RBC indices identified from our screening population, making a case for adoption of a universal thalassaemia screening strategy.

METHODOLOGY

We screened 10,409 patient samples in 2015 over a 1 year period. RBC indices were measured using Sysmex XE-5000. To qualify as normal, RBC indices had to fulfil the following; both MCV≥78fL and MCH≥27pg. Haemoglobin proteins were separated and identified using Sebia CAPILLARYS2 and Sebia HYDRASYS systems. Samples were stained with brilliant cresyl blue to detect HbH inclusion bodies.

RESULTS

9,352 cases (Figure 1) showed no abnormalities in this period, whereas haemoglobinopathies were identified in 1,057 cases (incidence of 10%).

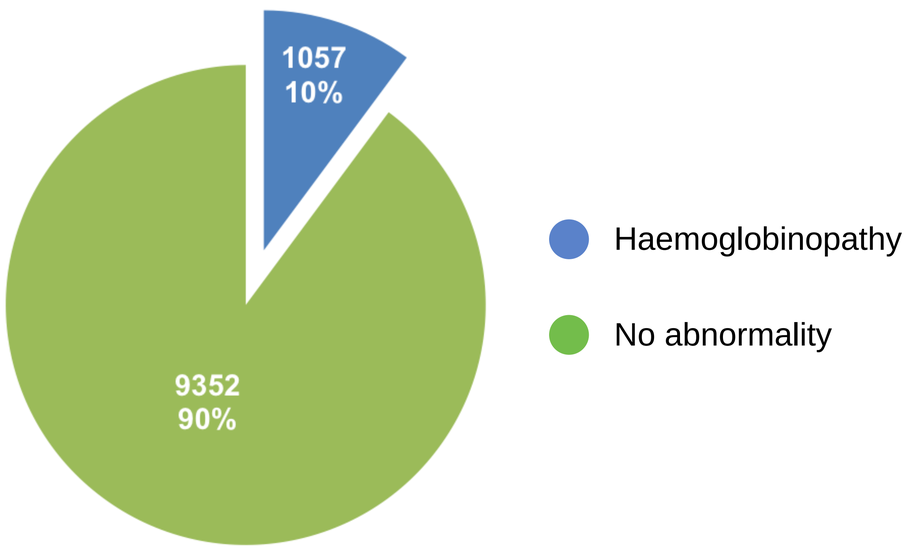

Figure 1. Among 10,409 patients screened in 2015, 1,057 haemoglobinopathy cases were identified.

Completely normal RBC indices were noted in 33 cases of these haemoglobinopathy cases: MCV (mean = 83.5fL, SD = 3.1fL); MCH (mean = 28.4pg, SD = 1.2pg); Hb (mean = 12.6g/dL, SD = 0.7g/dL for female; mean = 14.6g/dL, SD = 1.1g/dL for male). This constituted 3.1% of cases screened positive for haemoglobinopathies and 0.32% of the overall screening cohort.

Clinically significant haemoglobinopathies detected in these 33 patients (Figure 2) included Hb E trait (12 cases), Hb New York (6 cases), Hb Constant-Spring (5 cases), Hb S trait (3 cases), Hb J (3 cases), Hb D trait (2 cases), Hb Queens (1 case) and Hb Winnipeg (1 case).

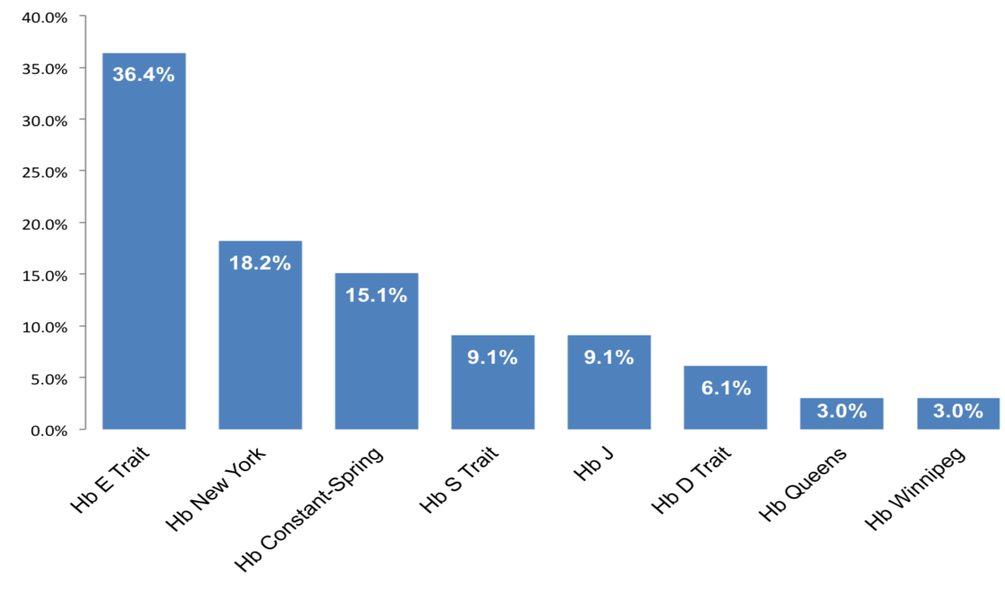

Figure 2. Proportion of haemoglobinopathy (in percentage) detected in 33 patients with completely normal RBC indices.

The proportions of patients presenting with normal or abnormal red blood cell indices for haemoglobinopathies detected are presented in Figure 3. While some haemoglobinopathies such as Hb E trait and Hb Constant-Spring manifest a high percentage of abnormal RBC indices, certain haemoglobinopathies such as Hb New York, Hb J, Hb Queens and Hb Winnipeg have a high percentage of normal RBC indices.

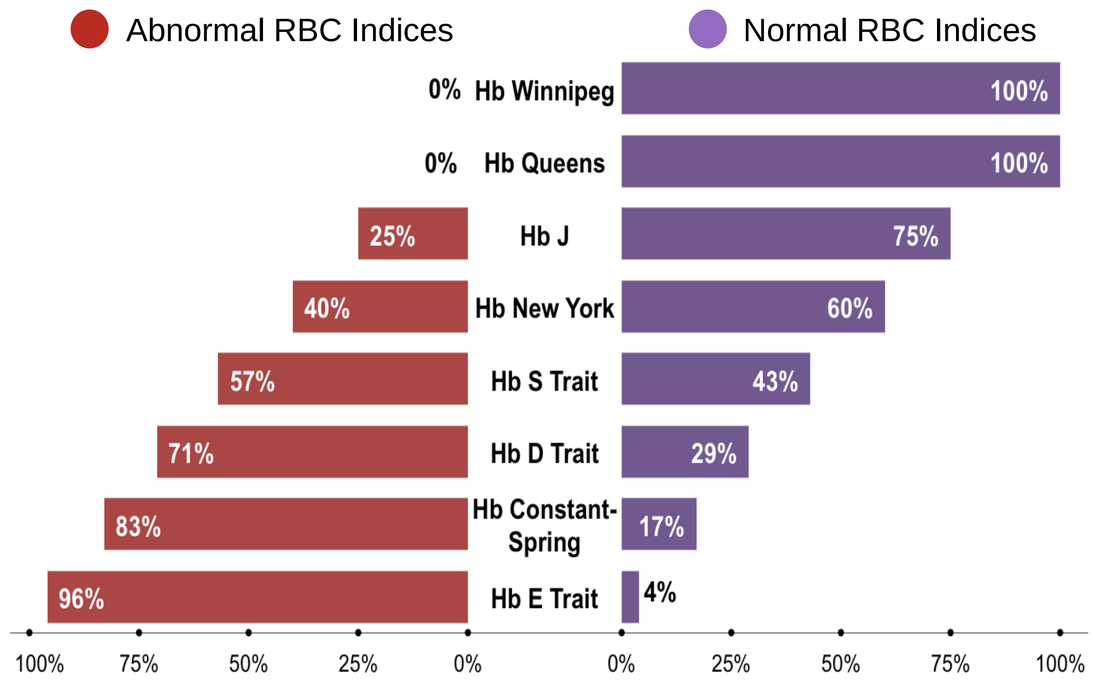

Figure 3. Comparison in percentage between normal and abnormal RBC indices within haemoglobinopathy classes.

CONCLUSION

Our data demonstrates that 3.1% of haemoglobinopathies identified from screening had completely normal RBC indices. This emphasizes the importance of universal thalassaemia screening to pick up asymptomatic carrier states during antenatal screening for accurate genetic counselling. This strategy will potentially help in further reducing the burden of major haemoglobinopathy disorders.