**P-224 - EXPANSION OF NEWBORN SCREENING IN QATAR TO INCLUDE SICKLE CELL, THALASSEMIAS AND OTHER HAEMOGLOBINOPATHIES**

Ramaswamy M, Skrinska VA, Mitri R, Hassan LR, Mohammed KAN, Abdoh G, AlMulla NAM, Badii R, AlSabbagh A

(1) Metabolic laboratory, Hamad Medical Corporation. (2) Newborn screening unit, Hamad Medical Corporation. (3) Paediatric Haematology, Hamad Medical Corporation. (4) Molecular genetics, HMC. (5) Haematology, HMC. Mamatha Ramaswamy - Doha, Qatar. mamadeep@gmail.com

**INTRODUCTION:** Haemoglobinopathies are a group of disorders which have abnormal production or structure of hemoglobin molecules. They are inherited as autosomal recessive. Qatar has a diverse population with more than 70% expats, predominantly from southeast Asian countries, middle east, Africa and Mediterranean countries. **OBJECTIVES AND METHODS:** Newborn screening for Sickle cell in Qatar was performed for two common mutations by molecular genetics. Since June 2018, we have expanded the screening to include thalassemias (α/β), Hemoglobin C/D/E using Bio-Rad VARIANTnbs in biochemical genetics. The new method uses high performance liquid chromatography. **RESULTS:** The switch of method from molecular to biochemical screen was done in collaboration of all the laboratory teams (genetics, biochemistry, hematology) with clinical teams (newborn screening unit and paediatric hematology). A total of 19,462 screens were performed on dried blood spots (DBS) from June 2018 to Jan 2019. During this 8 month period, we have identified 13 cases of sickle cell disease, 7 β thalassemia major, 5 hemoglobin D disease and 3 alpha thalassemia major. We also diagnosed 167 sickle cell traits, 90 Hemoglobin D traits, 35 Hemoglobin E traits and 18 Hemoglobin C traits. The confirmation is done by recall of the babies by newborn screening unit to collect whole blood for hemoglobin electrophoresis performed in hematology. The incidence of Sickle cell disease is 1 in 1500 livebirths (LB), β-thalassemia major 1 in 2800 LB, Hemoglobin D disease 1 in 3900LB and α-thalassemia major 1 in 6500LB. **CONCLUSIONS:** With the molecular screen, we were only able to detect sickle cell traits and sickle cell disease. The new method has helped us identify thalassemia major (α/β), Hemoglobin C/D/E traits and diseases which are highly prevalent in our population of Qatar. The limitation of new method is it cannot distinguish HbE from HbA2, as they produce peaks with same retention times; the latter is raised in β thalassemia. We are looking to introduce 2nd tier test on DBS using electrophoresis to distinguish the two peaks. This will avoid recall of the babies for collection of whole blood for electrophoresis. Also the method cannot identify Thalassemia traits (α/β) with sufficient sensitivity.