Reporting of Hemoglobin Traits: 11 Years of Experience from Maritime Newborn Screening

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Background: NBS services may report results not expected to affect health but important to the population. Maritime Newborn Screening (MNBS) tests for hemoglobinopathies and reports hemoglobin traits S, C, D, E, and unknown traits, but not suspected alpha thalassemia traits. The decision to report traits came from consultation with relevant patient populations/advocacy groups.

Methods: First tier testing for hemoglobinopathies is high-performance liquid chromatography (HPLC), and if abnormal the sample is tested via capillary electrophoresis (CE). Results are interpreted by a hematopathologist. Disclosure of results to families is done by a genetic counsellor (GC), and families are offered follow-up appointments with GCs for education, and testing if indicated. Data was gathered from laboratory case reports on positive screens for hemoglobin traits.

Results: Traits S, C, D, and E significantly increased from the first year of screening (total: 40) to the 11th (total: 205). There was a slight increase in unknown hemoglobin traits. During results disclosure families often share that they appreciate having the trait results and find this information helpful and reassuring. However, there are challenges in parental access to follow-up testing. Discussion: Reporting of unknown hemoglobin traits changed as MNBS gained experience in identifying variants confirmed by follow-up testing. MNBS initially reported suspected alpha thalassemia trait but discontinued this due to challenges with follow-up testing and workload. The drastic increase in S, C, D and E traits increased laboratory and clinic workload and prompted changes to improve service efficiency. The increase in traits is thought to corresponds with a shift in population potentially due to increased immigration. Conclusion: The majority of families receiving hemoglobin trait results appreciate having this information. Trait reporting by a GC provides an education opportunity for families and healthcare providers. MNBS plans to continue this service for hemoglobin S, C, D, E, and unknown traits.