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Title

Hemoglobinopathy screening and prevention in primary care in the Netherlands: informing further research within the US?

Priority 1 (Research Category)

Global Health

Presenters

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Abstract

Context

Hemoglobinopathies (HBPs), sickle cell disease (SCD) and alpha/beta thalassemia major, are the most common monogenic autosomal-recessive diseases in humans. Carriers are generally asymptomatic and unaware of their carrier status. We present two completed studies on Hemoglobinopathy screening and prevention in The Netherlands, informing research in progress in the Mayo Clinic Health System.

Objectives

The aim of both studies was to explore better strategies for HBP prevention in the preconception and antenatal phase in primary care practice (PCP).

Study Design and Analysis

1) Prospective quantitative study was carried out in The Hague region from different data sets (Hemoglobinopathies Reference Lab of the Leiden Diagnostic Genome Analysis laboratory (LDGA) at Leiden University Medical Center (LUMC), the Extramural LUMC Academic Network (ELAN) and the HBP patient database at the HAGA hospital in The Hague). To calculate prevalence, we used the HAGA database containing all patients with beta-thalassemia intermedia/major and sickle cell disease in The Hague region. Carrier prevalence was calculated using the information provided by the Hemoglobinopathies Reference Lab within the LDGA at the Clinical Genetics department of the LUMC and the ELAN database.

2) A qualitative study was used (interviews of 10 HBP patients, 10 carriers and 10 PCPs) to explore knowledge about HBP and the need for education and communication in those areas. The interviews were audio-recorded, transcribed verbatim and analysed using content analysis to identify recurring themes (using software ATLAS.ti).

Results

1) Highest prevalence of hemoglobinopathies in immigrant neighborhoods with large gap between estimated carrier prevalence and the actual registration of carriers in electronic patient records.

2) Three themes were identified:(1) a need for more information on HbP, (2) a need for indications when to refer for analysis (carrier diagnostics) and (3) insight concerning organization and roles in care for hemoglobinopathy carriers and patients, reflecting a need to increase awareness, improve competences among PCPs (education, communication with patients and their unidentified family members).

Conclusions

Both studies showed a critical need for action to improve informed reproductive decision making for the at-risk population and more research is needed to define the requirements of US and European healthcare system to address this challenge.