

HOW ICT MIGHT SUPPORT ACCESS TO CLINICAL EXPERTISE FOR TIMELY DIAGNOSIS OF GENETIC DISORDERS IN IRISH TRAVELLERS AT POINT OF CARE

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ABSTRACT

Irish Travellers have high levels of consanguinity, resulting in many rare genetic disorders. Most of these disorders are as a result of specific mutations unique to this population. Clinicians who are less familiar with this population have few resources to facilitate forming a differential diagnosis or in targeting genetic testing. For people with a rare disease, the mean average length of time from symptom onset to accurate diagnosis is approximately 4.8 years. 40% of rare disease patients are misdiagnosed at least once. The longer it takes to diagnose a rare disease, the more health care professionals the patient needs to see. This problematic journey to diagnosis and care can increase medical, economic and social burdens. An expert group of clinicians have recently collated the disorders to facilitate a targeted genetic approach to diagnostics in Irish Travellers.

The literature review demonstrates the availability of web-based ICT for the diagnosis of genetic disease in specific ethnic groups in other countries. A motive for this study is to support more timely diagnosis of these rare disorders in Irish Travellers by the introduction of a similar web-based initiative. Accordingly, the first aim of this study was to determine how a similar diagnostic support resource for genetic disease in Irish Travellers might look.

The barriers to timely diagnosis of rare genetic disease in Irish Traveller patients are explored. A gap in the literature established that there is scant material available relating to the means by which Traveller ethnicity is reliably captured in paper charts or electronic health records (EPR), thereby hindering implementation of a similar diagnostic support initiative for the Irish Travellers. The Beutler test was identified as an enhanced newborn screening process for all Traveller infants and was subsequently used as a starting point to explore the current process for capture of Traveller ethnicity in health records and EPR. The second project aim was to explore how Traveller ethnicity could be employed to trigger the use of the web resource at an appropriate point in the diagnostic workflow.

A qualitative method was devised to explore local expert knowledge to fill gaps identified in the literature. Thirteen domain experts were identified and chosen as participants for a semi-structured interview. Each question set was tailored to the participant, and interviews roughly fell into three categories: interviews to inform prototype, interviews to describe the Beutler process, and interviews to describe diagnostic workflows and order communications.

Literature review and qualitative analysis of interview findings facilitated construction of narrative descriptions of Traveller ethnicity capture in Irish health records, Traveller ethnicity as a trigger for the Beutler test and options for the creation of a Traveller flag in electronic health records. The means by which Traveller ethnicity is currently captured in Irish health records and EPR was defined. The enablers and obstacles to reliable identification of Travellers within national ICT projects are described. Ethical and legislative barriers are discussed.

Information gathered was used to inform prototype design of a web resource for the diagnosis of genetic disorders in Irish Travellers. A point of care for intervention, within the limitations of national ICT projects, is identified.