

Utilization of Genetic Resources in Pediatric Emergency Medicine

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Genetic disease, particularly when undiagnosed and untreated, can greatly impact childhood health. Children with genetic disease present more frequently to the emergency department for health concerns (Kumar, Radhakrishnan, Chowdhary, & Giampietro, 2001); however, to our knowledge, there is no data on the practices and perspectives of emergency physicians regarding identification of pediatric patients at risk for genetic disease. In this study, 180 pediatric emergency physicians were surveyed about their referral practices, perspectives, recommendations, and interest in increasing utilization of genetic resources in pediatric emergency medicine. This study found that while 89% of respondents have access to a local genetics provider and 89% believe it is their role to refer patients for genetic evaluation (either directly or through a primary care physician), 35% have never recommended or referred a patient for genetic evaluation in their career in pediatric emergency medicine. Barriers to identifying genetic disease and utilizing genetics resources reported in this study were: lack of relevance to the immediate treatment at hand, lack of time, and lack of genetics training for emergency providers. Resources desired by study participants included increased genetics training, protocols for referral practices, and access to genetics providers. Despite these barriers, 81% of surveyed physicians reported interest in increasing the utilization of genetic resources in their clinical practice. Study findings identify a need for increased genetics training for pediatric emergency physicians along with improved access to genetics providers.