Implementation of an automated prediction scoring system to identify patients at possible increased risk for Hereditary Angioedema

Marissa Shams, MD 1, Dawn A. Laney, MS *1 ,Dave A. Jacob, BS 2 , Jingjing Yang, PhD 1,
Jessica Dronen, MS 3 , Amanda Logue, MD 4 , Ami Rosen, MS 1 , Marc Riedl, MD 5

1 Emory School of Medicine
2 ThinkGenetic Foundation
3 ThinkGenetic, Inc.
4 Lafayette General Health / Ochsner
5 UC San Diego Health

Poster number 124

Background

Hereditary angioedema (HAE) is a genetic condition characterized by intermittent and recurrent episodes of severe swelling (angioedema). Swelling can be life-threatening as it can occur unexpectedly. Patient identification is important as there are several FDA approved therapies that can reduce the frequency and severity of painful edema events. Many individuals still experience long delays to diagnosis. A pilot study in HAE was undertaken to determine if patients who may be at risk can be flagged from de-identified medical records data fields.

Hypothesis

The development of an automated "early warning system" that identifies patients at risk from medical record data can be integrated into standard medical appointment workflow and educate specific physicians about HAE in a "just in time" methodology that will decrease the time to diagnosis and treatment as compared to published numbers on average time to diagnosis and treatment.

Methods

A prediction scoring system for HAE was created and validated using known cases of HAE from the medical literature as well as positive and negative controls from HAE-focused centers. Using key features of medical and family history, a series of logistic regression models for the five known genetic causes of HAE were created. Top variables populated the digital suspicion scoring system and were run against de-identified electronic health record (EHR) data. Patients were categorized as increased, possible, or no increased risk of HAE at two diverse sites.

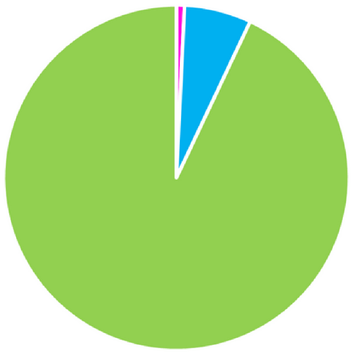

Figure 1. Risk Level Distribution

- High (200+) 0.15%
- Possible (100-199) 6.31%
- No Increase (0-99) 93.54%

Prediction scoring using the strongest 13 variables on the “real world” EHR positive control data identified all but one C1-inhibitor deficiency case and one non-C1-inhibitor deficiency case without false positives. The two missed cases had no documented family history of HAE in their EHR. When the prediction scoring variables were expanded to 25 variables the screening algorithm approached 100% sensitivity/specificity. The 25 variable algorithm run on general population EHR data identified 26 patients as increased risk for HAE at the medical centers.

Development, validation, and implementation of automated prediction scoring systems can be useful to aid providers in identifying patients with rare genetic conditions.

Limitations

Demographic information was not used to pre-screen patients included in this pilot study. All patients presenting at participating facilities during the study time period had equal opportunity to be included in analysis. However, this may not be a true representation of the area and may not generalize to other geographic locations. Due to the inability for follow-up in this pilot project we are not able to determine the clinical specificity and sensitivity of these algorithms at a particular facility.

ThinkGenetic Foundation
ThinkGenetic
Ochsner Lafayette General
EMORY UNIVERSITY
UC San Diego

Acknowledgements

This work was supported in part by an investigator initiated study grant funding from Takeda to the ThinkGenetic Foundation. We would also like to thank our colleagues at LGH, Guardian Research Network, consulted experts in the field, our statistician colleagues, and our dedicated technical development team for their contributions to this project.