

See discussions, stats, and author profiles for this publication at: <https://www.researchgate.net/publication/314235544>

Almost Zero Error Basepair-based Record Alert (AZE BRA) – A Genomic Clinical Decision Support Tool (ABSTRACT)

Poster · March 2017

DOI: 10.13140/RG.2.2.12032.64008

CITATIONS

0

READS

10

4 authors, including:



Anand Kulanthaivel

Indiana University-Purdue University Indianapolis

6 PUBLICATIONS 4 CITATIONS

SEE PROFILE

Some of the authors of this publication are also working on these related projects:



Almost Zero Error Basepair-based Record Alert (AZE BRA) – A Genomic Clinical Decision Support Tool

[View project](#)

Almost Zero Error Basepair-based Record Alert (AZE BRA) – A Genomic Clinical Decision Support Tool

Anand Kulanthaivel, M.I.S.¹, Madhura Kshirsagar, M.S.¹ Mohammad Alarifi, M.S.²,
Mark Oklak, M.S.¹, Josette F. Jones, R.N., Ph.D.¹

¹Indiana University – Purdue University Indianapolis, Indianapolis, IN, USA

²University of Wisconsin-Milwaukee, Milwaukee, WI, USA

Introduction & Background

The idea of the United States's Precision Medicine Initiative (PMI)¹ was to allow providers (and patients) to leverage large amounts of information (including patient genomic data)² in order to create actionable knowledge that increases patient well-being. To this end, we propose a system called AZEBRA; the acronym stands for Almost Zero Error Basepair-based Record Alerts. (*Zebra*, in addition to being a well-known wild animal, is a common medical slang term for the clinician's fallacy of mistakenly coming to a rare and sometimes dire diagnosis (the rare *zebra* diagnosis) due to having missed more common causes of patient symptoms (the common *horse* diagnosis); conversely, patients with rare conditions would be better thought of as zebras and not horses.³) AZEBRA is intended to leverage the principles of genetically-enhanced precision medicine in order to alert clinicians to the presence of patients with one common and four rare genetic pathologies that are ordinarily sources of unnecessary morbidity and mortality in clinical settings.

Clinical Knowledge; Technical Brief

Knowledge content to develop the actual logic modules for AZEBRA was taken from PubMed's Clinical Variant (ClinVar) SNP database. Four rare (as classified by the National Organization for Rare Disorders) disorder pathologies, Marfan Syndrome, Ehlers-Danlos Syndrome, Acromicric Dysplasia, and Osteogenesis Imperfecta, as well as one more common pharmacogenomic pathology (Cytochrome P450-2D6 Deficiency) are used as test cases for AZEBRA. With respect to these 5 disorder pathologies, 74 known associated significant pathological genetic variants (as encoded by single nucleotide polymorphisms; SNPs) were recorded in the ClinVar database, and a set of alerts and contextual conditions upon when they will be launched was generated and placed into our data model.

The model we are developing for the system is thus compatible with Health Level 7 Version 3 (HL7v3) storage and retrieval via Fast Healthcare Interoperability Resources (FHIR); this type of model is more likely to be compatible with modern Electronic Medical Record (EMR) systems.⁴ For storage of the data, we believe that a star-schema data warehouse model is most appropriate due to our expectations of high query volume combined with relatively low input rates. The system begins with an invocation (clinical situation) and then checks to see if the patient has potentially hazardous pathologies (as matched to patient genetic variants through the fact table) that match the ones targeted by the given invocation. The appropriate message is then generated.

Conclusion; Future Work & Needs

It is expected that the resulting system, if implemented, will reduce mortality and morbidity due to undetected pathological genomic variants and will be the first of its kind to function directly as a decision support aid in the clinic. Future work (some of which will be accomplished by time of presentation) will include: the actual construction, integration and loading of the required information model; and the full development of the logic of each of the five identified use cases. Work beyond that phase will include the usability testing of the interface and logic of the system with volunteer clinicians, system refinement, and the addition of further biomedical knowledge to the system's logic. Finally, we acknowledge that the education of clinicians and patients to the benefits of proper genetic testing and tailored alerts must occur in order for the system to be adopted.

References

1. The Whitehouse (United States of America). *Fact sheet: President Obama's Precision Medicine Initiative*. (2015). Retrieved from <https://www.whitehouse.gov/the-press-office/2015/01/30/fact-sheet-president-obama-s-precision-medicine-initiative>
2. Aronson, S., Rehm, H.L.). Building the foundation for genomics in precision medicine. *Nature*. 2015;526(7573): 336-342.
3. Sotos, J., *Zebra Cards: An Aid to Obscure Diagnoses*. 2006, Mt. Vernon, VA: Mt. Vernon Book Systems.
4. Health Level 7. Fast Healthcare Interoperability Resources (FHIR). (2002-2016). Retrieved from <http://hl7.org/fhir>.