Realizing the Value of Genomic Decision Support in Healthcare Systems: *Opportunities, Challenges, and Strategies for Success*

Chunhua Weng, Columbia University

Physician Knowledge Gaps
(Informational Needs)

What genetic syndrome should I suspect in this patient?

What genetic test should I order?

How do I interpret the genetic findings?

What are the next steps in management?

When should I refer the patient?

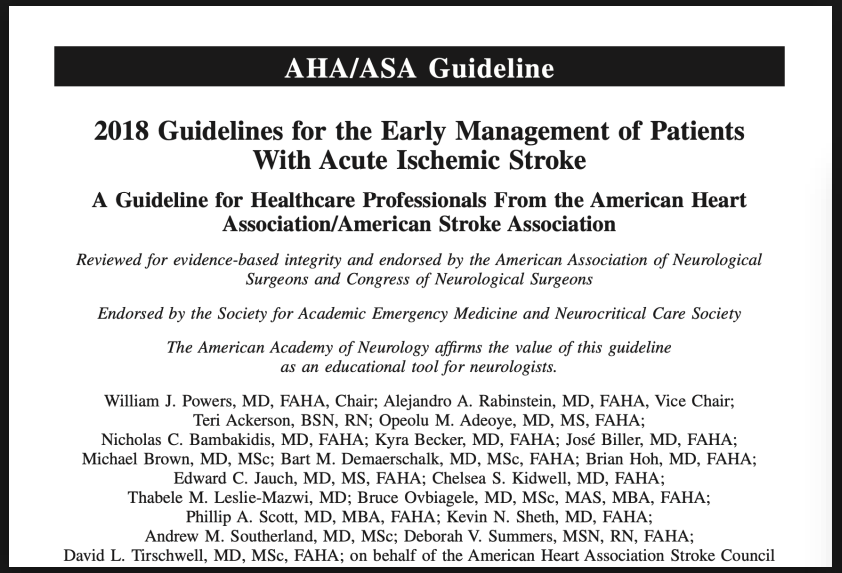
Who else in the family should get testing?

Where can I find management guidelines?

Jordan Nestor, MD

Clinical Decision Support (CDS) tools for clinicians lacking genomic expertise

I. Select FH management guidelines and convert text into computer-interpretable modules



AHA/ASA Guideline

2018 Guidelines for the Early Management of Patients With Acute Ischemic Stroke

A Guideline for Healthcare Professionals From the American Heart Association/American Stroke Association

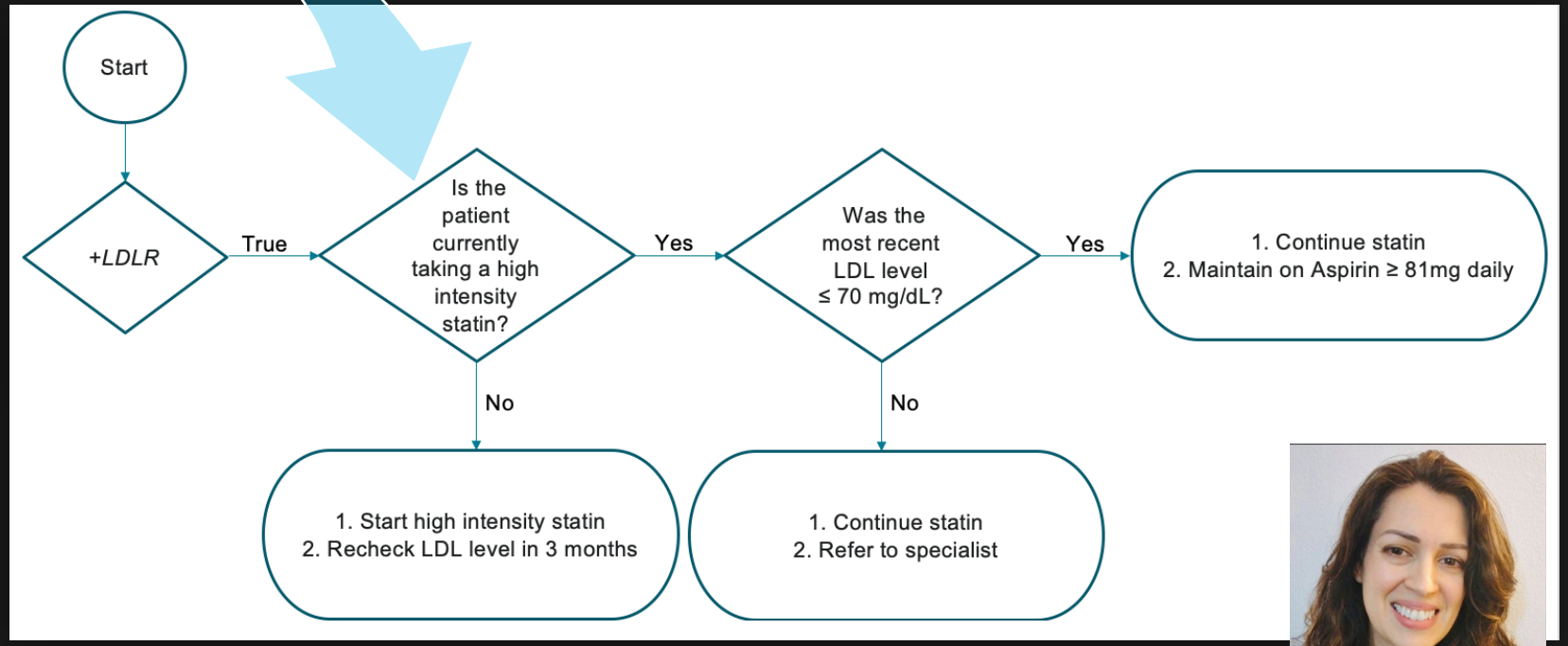
Reviewed for evidence-based integrity and endorsed by the American Association of Neurological Surgeons and Congress of Neurological Surgeons

Endorsed by the Society for Academic Emergency Medicine and Neurocritical Care Society

The American Academy of Neurology affirms the value of this guideline as an educational tool for neurologists.

William J. Powers, MD, FAHA, Chair; Alejandro A. Rabinstein, MD, FAHA, Vice Chair; Teri Ackerson, BSN, RN; Opeolu M. Adeoye, MD, MS, FAHA; Nicholas C. Bambakidis, MD, FAHA; Kyra Becker, MD, FAHA; José Biller, MD, FAHA; Michael Brown, MD, MSc; Bart M. Demaerschalk, MD, MSc, FAHA; Brian Hoh, MD, FAHA; Edward C. Jauch, MD, MS, FAHA; Chelsea S. Kidwell, MD, FAHA; Thabele M. Leslie-Mazwi, MD; Bruce Ovbiagele, MD, MSc, MAS, MBA, FAHA; Phillip A. Scott, MD, MBA, FAHA; Kevin N. Sheth, MD, FAHA; Andrew M. Southerland, MD, MSc; Deborah V. Summers, MSN, RN, FAHA; David L. Tirschwell, MD, MSc, FAHA; on behalf of the American Heart Association Stroke Council

Decision Tree Modeling



Develop a Genome-informed CDS tool for Familial Hypercholesterolemia (FH)

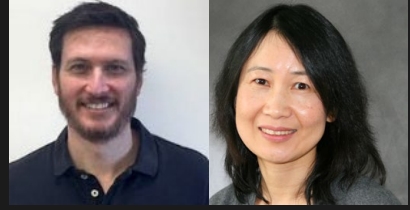
Jordan Nestor, MD

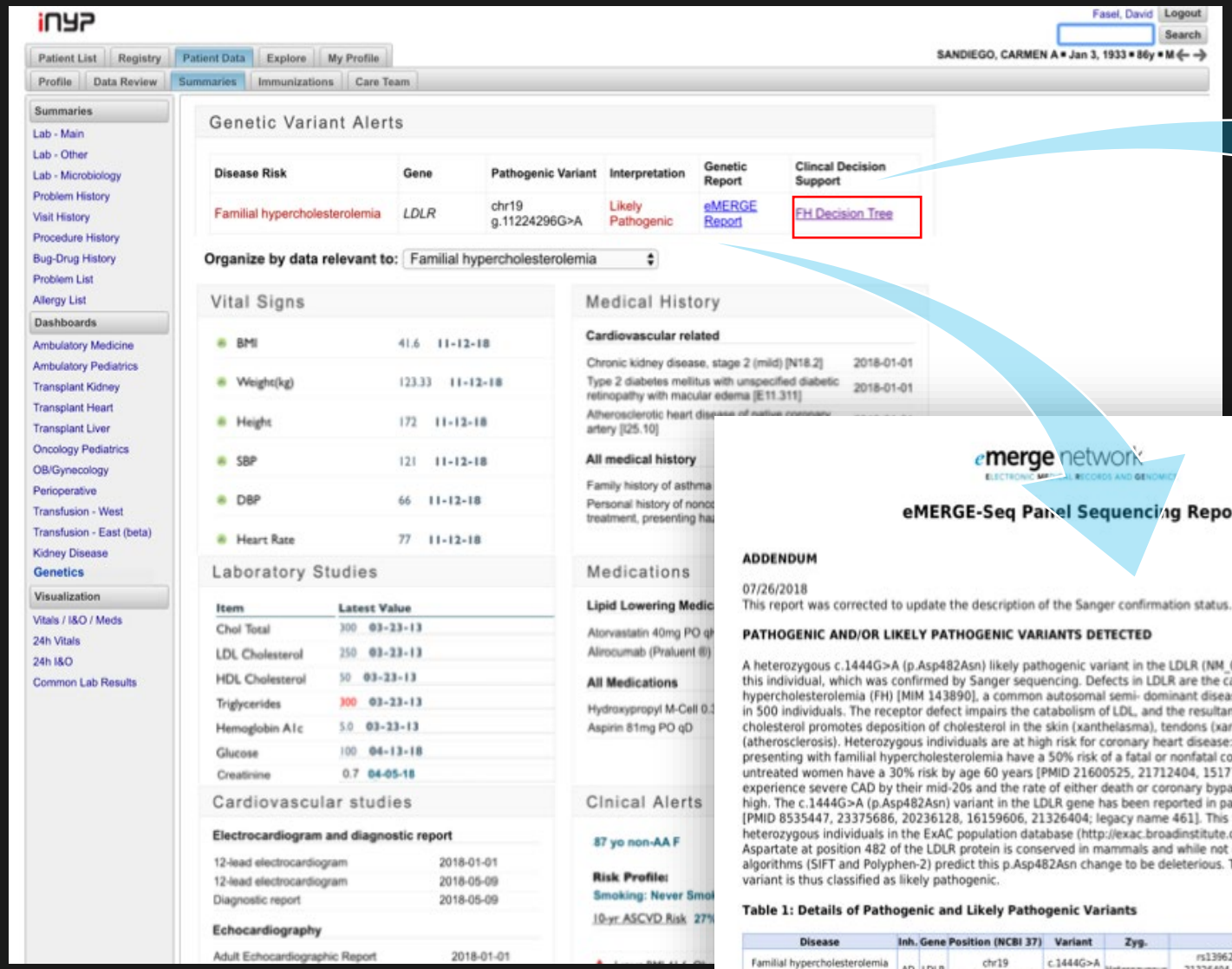
II. Conducted a scenario-based evaluations of decision tree's logic among target users and assessed ease of use and usefulness in clinical practice

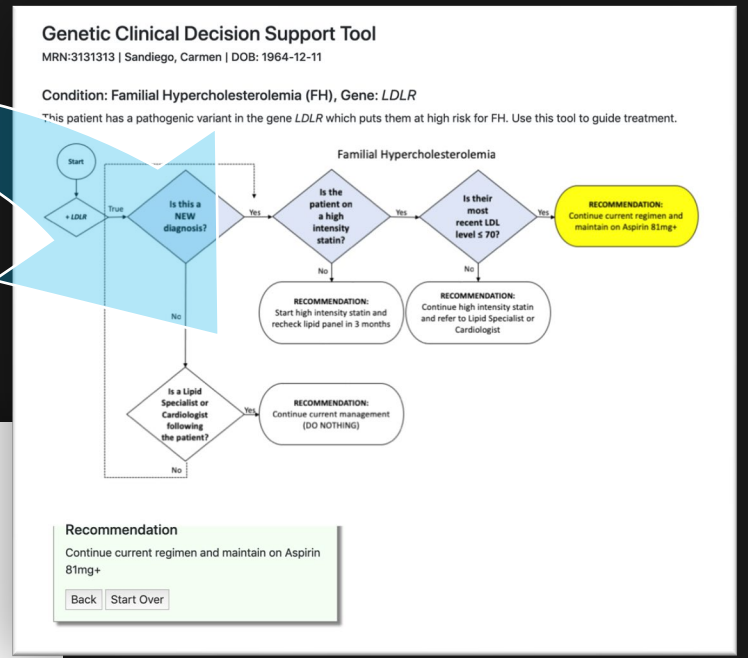
User	Unmet Information Needs
1	"I think you should also list the different high intensity statins"
2	"I'd like to see a link to the full guidelines in the tool"
3	"This doesn't work when the LDL is already less than 70. The user isn't then prompted to start a high intensity statin"
4	"Is genetic testing diagnostic for this condition?...The diagnostic criteria need to be shown here because I don't know what it is… and the flowchart makes no mention that this is diagnostic for FH"
5	"The first branch point does not give enough information" "I'm unclear as to when I should refer to a specialist" "I didn't know what the LDL goal is supposed to be unless they're already on a high intensity statin"
6	"I'd just to have to know more information about their cardiovascular risk before being able to feel comfortable managing the patient on my own"

Jordan Nestor, MD

III. Made iterative changes and then implemented the prototype into the EHR as a proof of concept







Challenges

Solutions & Tradeoffs

Competing Guidelines → **Merge multiple guidelines**

Ongoing barriers to integrating raw genomic data in EHRs → **Upload PDF files of genetic test results**

Heterogenous IT Infrastructures → **Create an interactive web-based decision tree within a customized EHR dashboard**

Do clinicians use unsolicited genomic data integrated into the EHR?

Jordan Nestor, MD

Only 1% of all the eMERGE-3 genetic test results in the EHR were viewed by clinicians, who did not initiate genetic tests

Jordan Nestor, MD

Characteristics	Clinician Users *N*=30
Attendings	**14 (47%)**
Fellows	4 (13%)
Residents	9 (30%)
Nurse practitioners	3 (10%)
Anesthesiology	4 (13%)
Surgery	3 (9%)
Radiology	2 (6%)
Neurology	1 (3%)
Pathology	1 (3%)
Medicine	**19 (63%)**
During "regular" workhours†	**23 (77%)**
During "on-call" hours†	6 (20%)
During "regular" and "on-call" hours	1 (3%)
Outpatient/Ambulatory	**18 (60%)**
Inpatient/Hospitalized	11 (37%)
Both outpatient and inpatient settings	1 (3%)
Around the time of a scheduled outpatient clinical encounter	11 (37%)
Care team member of a hospitalized participants	11 (37%)
Established outpatient provider	5 (17%)
Not ascertained/Unknown	3 (10%)

Key Barriers

1. Disconnect between genomic discovery (by scientists) and healthcare delivery (by clinicians)
2. Lack of coordination, workflow support, and policy for liability at hospitals
3. Lack of current knowledge to practice evidence-based genomic medicine among clinicians
4. Poor EHR user interface (hard to make things found), alert fatigue, information fragmentation,
5. No genomic knowledgebase (standards-based, computable, reasonable, provenance checking
6. Seamless integration between genomic knowledge base and current PubMed literature
7. Integration of the knowledge to EHR for automated triggering of genomic practice guidelines

Enablers

1. Opportunities: Learning health system development within CTSA hubs
2. EHR-based informatics research results + implementation science
3. Decades of clinical informatics research on knowledge representation and reasoning
4. Text mining and evidence inference
5. FHIR and other EHR data exchange standards

Sociotechnical Strategies for Success

1. Invest in informatics research for genomic evidence computing and genomic knowledgebase construction to enable scalable, sharable and computable inferences of genomic knowledge and harmonization of practice guidelines
2. Harmonize the interests of multiple stakeholders (EHR venders, Hospital IT, genomic scientist, clinicians, and institutional leadership) to facilitate team science and implementation science
3. Forge collaborations to develop a learning health system for genomics (what is new evidence, who is affected, what needs to be done, etc)
4. Clinician-centered design of workflow support, referral support, CDS, etc.

Thank you!