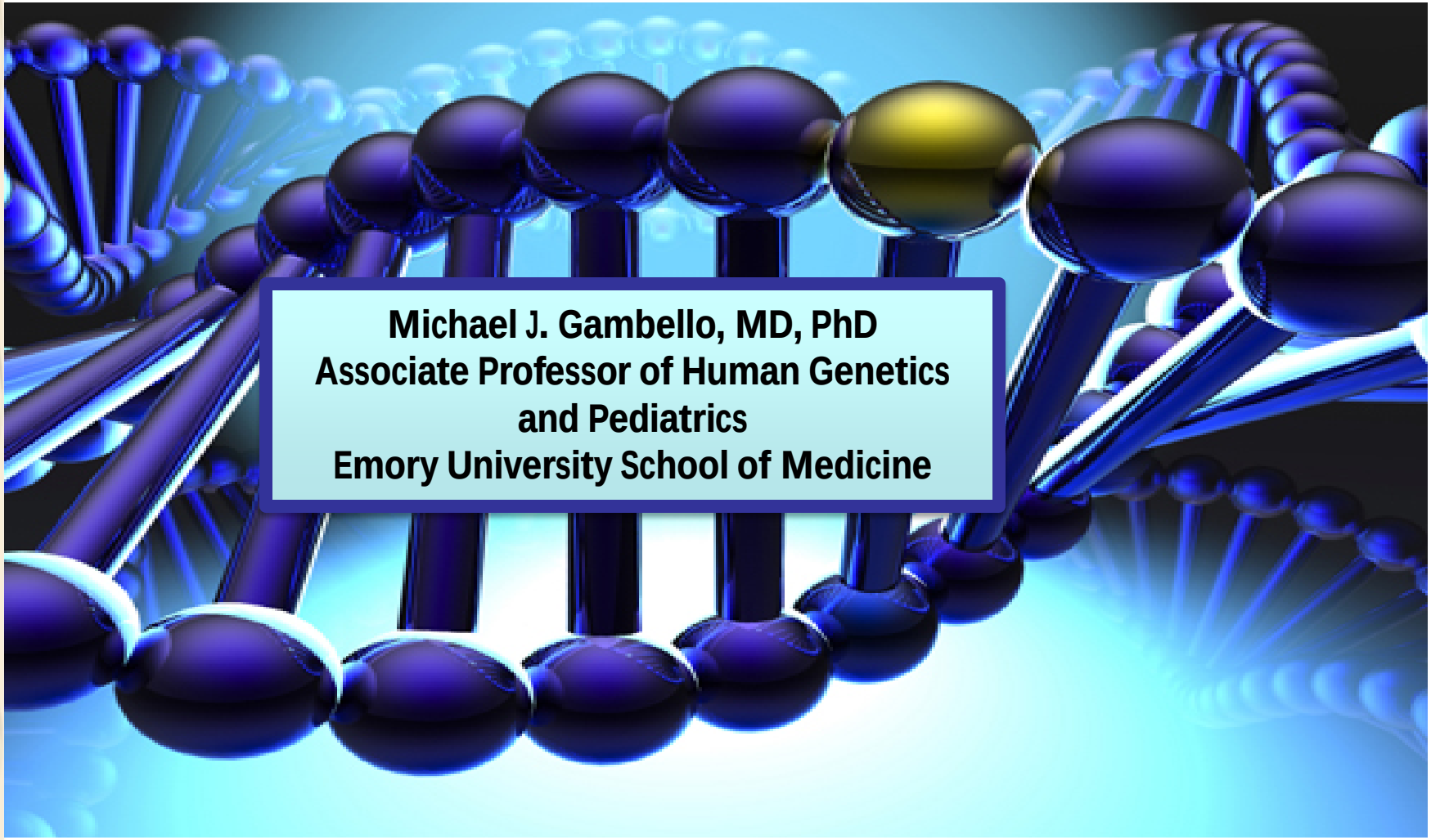
EMORY UNIVERSITY SCHOOL OF MEDICINE

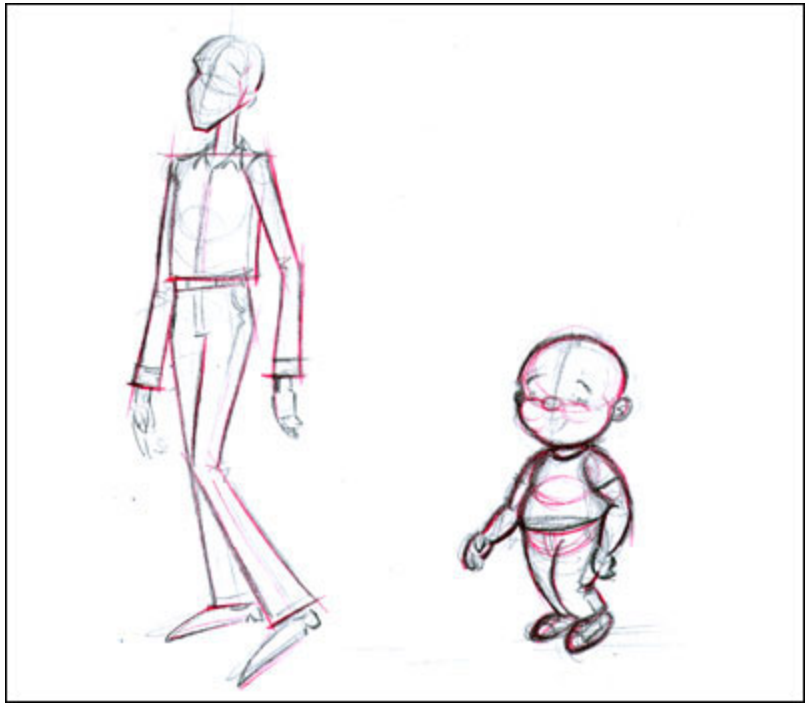
The Department of HUMAN GENETICS

Translating Genomic-Based Research for Health Care Decision-Making



Michael J. Gambello, MD, PhD
Associate Professor of Human Genetics and Pediatrics
Emory University School of Medicine

Medical Genetics Clinic



15% Adult 85% Pediatric

- 30 new patients/week
- 1500 visits/year
- Rare Diseases
- Developmental Delay
- Autism
- Family history of genetic disease

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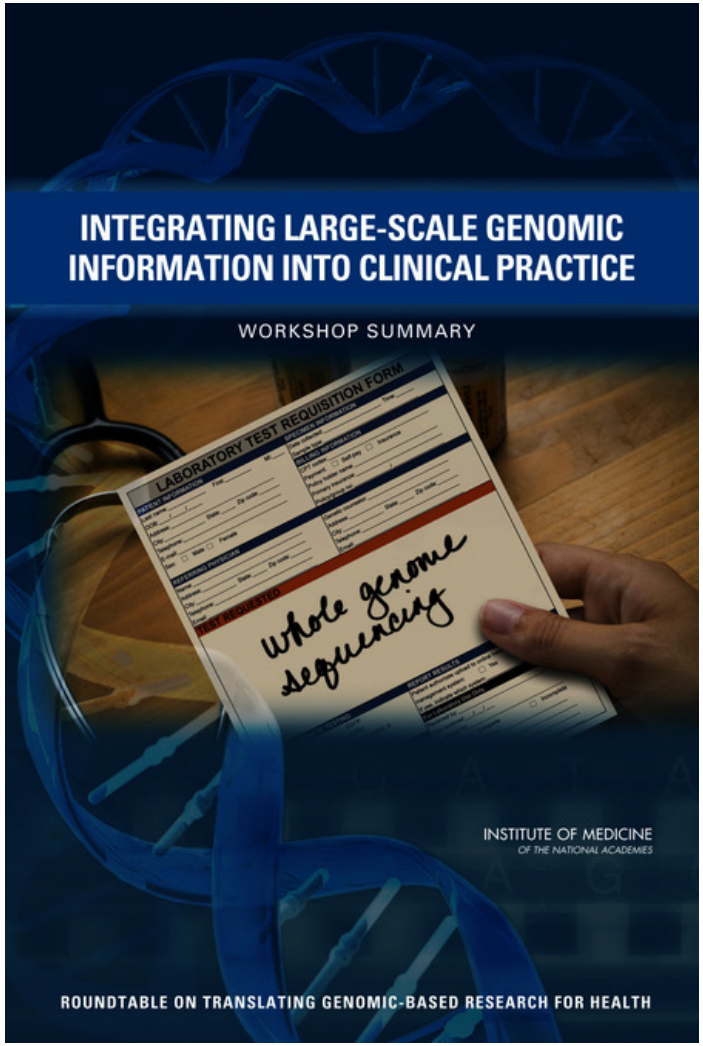
Rare Disease and Genomic Medicine



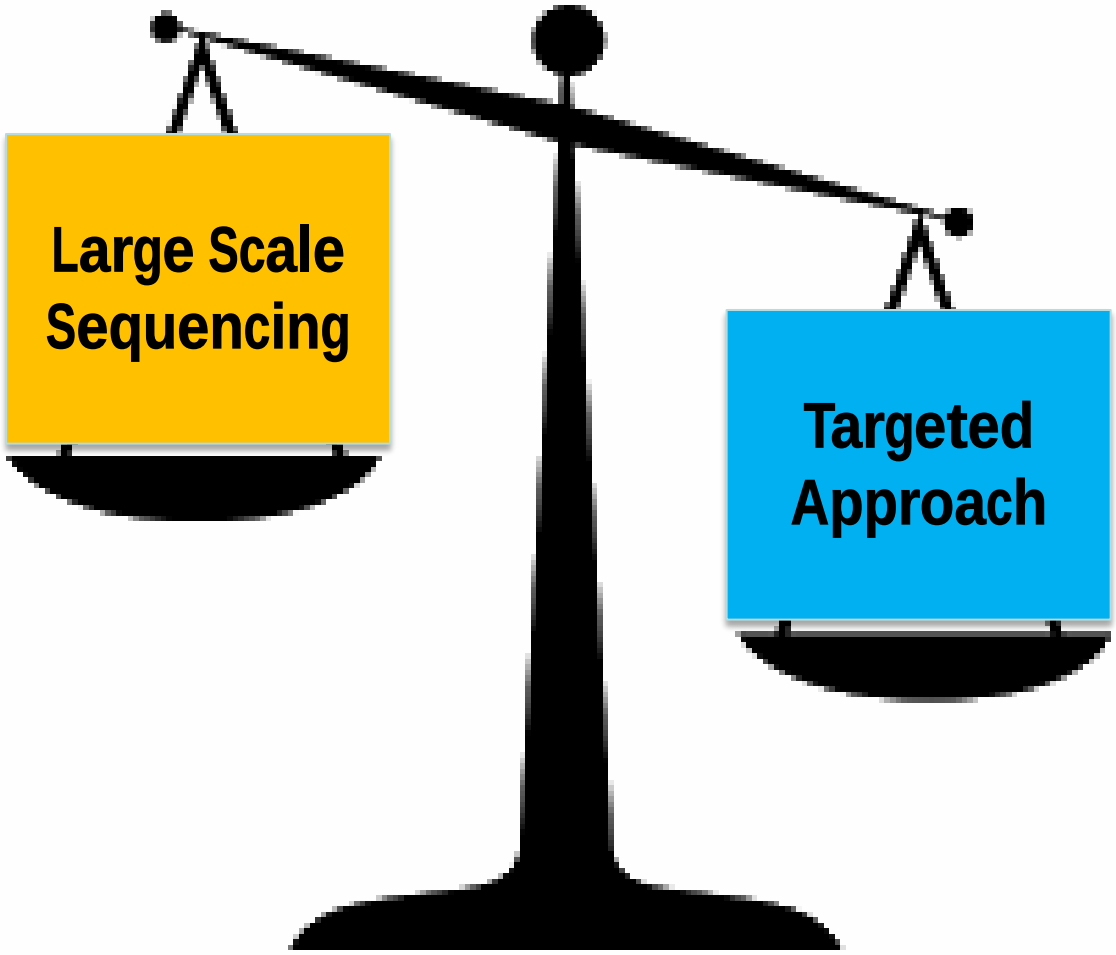
The study of rare diseases offers a way of implementing the tools and procedures that will later be used in more widespread applications of genomic medicine.

What's wrong with my child (me)?
What caused it?
What can be done about It

Les Biesecker, MD

Genomic Medicine?

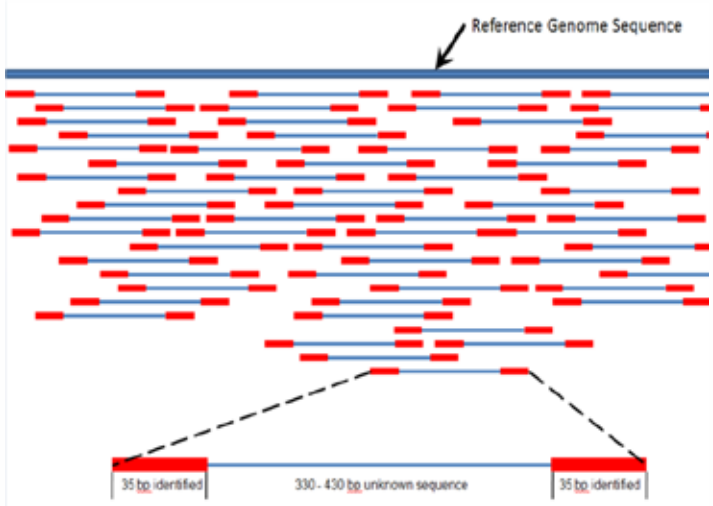
VS.



- Targeted Approach = Good Medicine
- History and PE lead to a Differential Diagnosis
- Tests that are likely to make a diagnosis.
- Very important when there is a recognizable genetic disorder.
- Large scale sequencing cannot diagnose all genetic disorders.

When to Use Genomic Medicine?

Large Scale Sequencing



ACMG POLICY STATEMENT | Genetics inMedicine

GENETICS in MEDICINE | Volume 14 | Number 8 | August 2012

a) Likely Genetic, but no specific genetic test available.

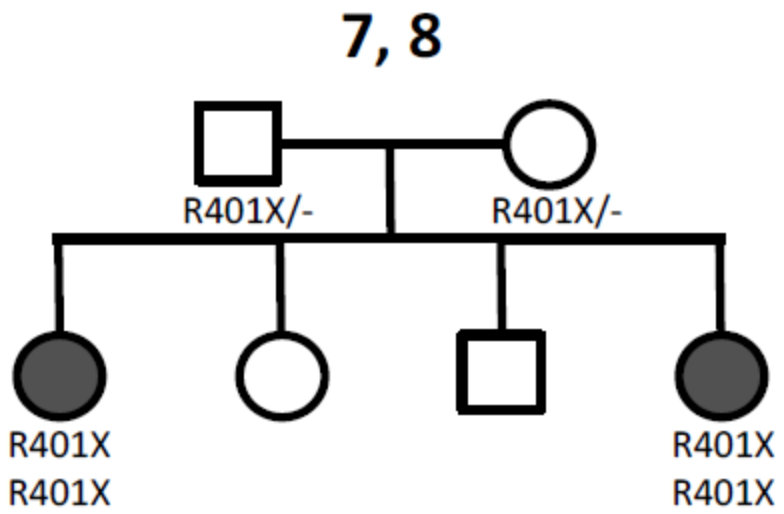
b) Genetic Disorder, but so many genes involved, better to test many.

c) Likely Genetic, but targeted tests have Not yielded a diagnosis.

d) Fetus with likely genetic disorder, but targeted tests have not yielded a diagnosis

Discovery of NGLY1 Disease

c) Likely Genetic, but targeted tests have Not yielded a diagnosis.



Other Genomic Indications

b) Genetic Disorder, but so many genes involved, better to test many at once.

Cardiomyopathy Panel
Aortic Aneurysm Panel
Childhood Epilepsy Panel
Retinal Disease Panel
Neuromuscular Panel

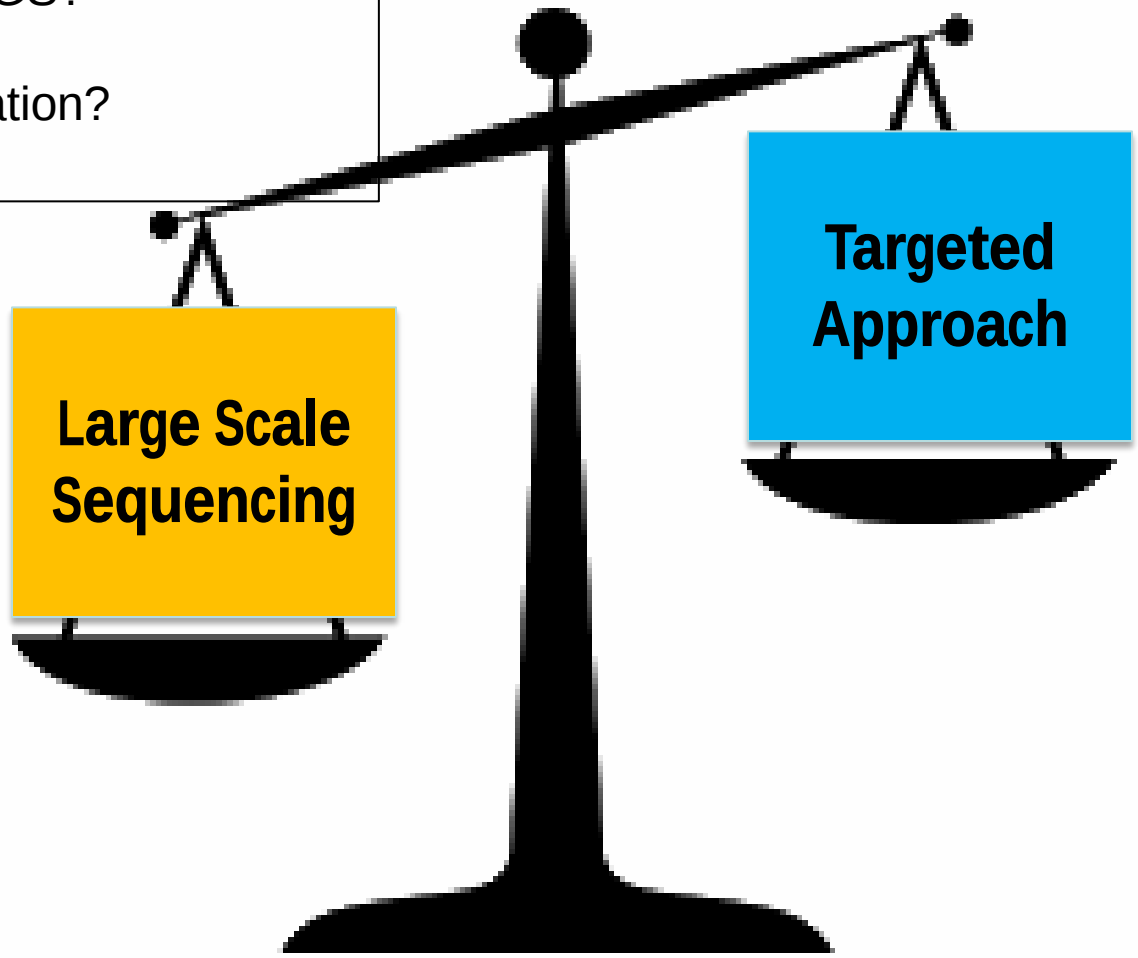
Targeted vs Large Scale???

Cost?
How much Targeted Approach
Until WES/WGS?

Clinical Indication?
Fetal Demise

?

Will Genomics affect Clinical Reasoning
An exome for Down syndrome???



Limitations

- **Does reimbursement play a role in ordering a whole genome test?**

ABSOLUTELY

Limitations

Georgia Medicaid (60%):

Zero reimbursement for Whole Exome Sequencing.

Inpatient Exome:

Reviewed by Pathology.

If not required for acute care, often denied.

Patient Deductible

Coping with Limitations

- Some clinical labs have lower financial thresholds than others
- Research Funds
- Mendelian Center (Baylor/Johns Hopkins)
- Wait until prices decrease further
- Philanthropic support

Patients Preferences

- Limited role in what testing is done.
- Some parents push more for diagnoses.
- Lab consent forms very important and helpful.
- Almost all patients want all information.
- Not had to deal with incidental findings yet.

Patient Understanding

- Extremely dependent of patient education level.
- Patients (Parents) tend to want ALL the information.
- Not always clear how much they understand the implications of incidental findings.

Summary

- Evaluation of Rare Diseases will continue to translate Genomics to Patient Care.
- Clinicians will need to continue to evaluate Targeted vs Large Scale Sequencing.
- Financial considerations.
- Patient understanding, needs, care.
- Evidence based recommendations always in demand.