Talking Genomes with Your Patients

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Objectives

- Review the importance of physician familiarity with genomic testing and results
- Explore common patient (and physician) misconceptions and re-education techniques
- Discuss strategies for pre- and post-test conversations with your patients
- Identify resources for genetic and genomic information for patients and providers

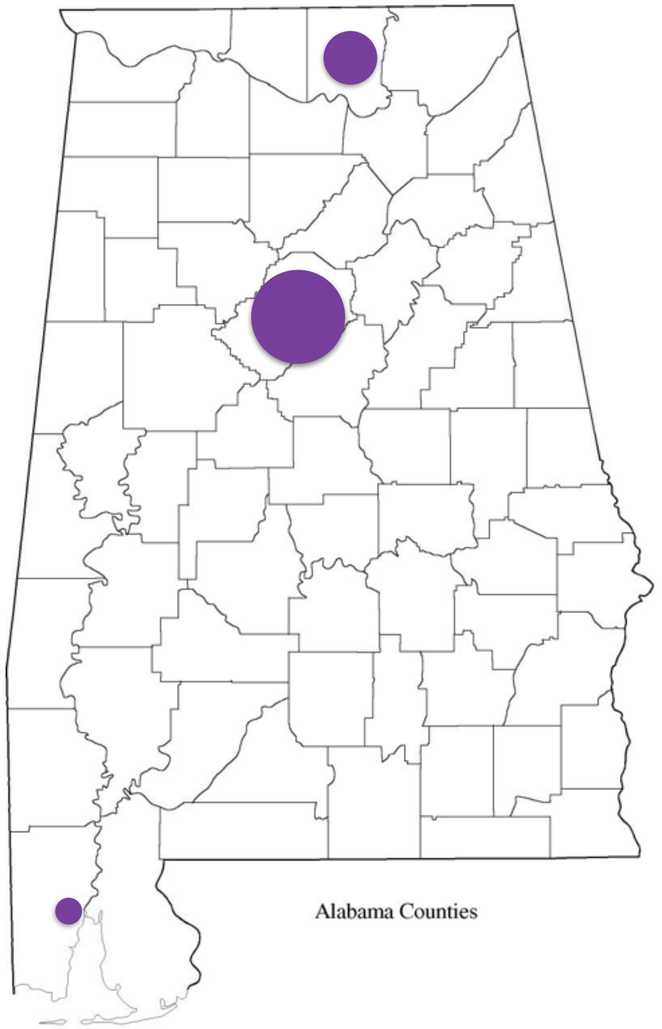
Why Does This Matter?

You may be the sole source of genetic information for your patients

There are not enough genetic counselors/ geneticists to handle every patient or every test that needs to be done

Genetics/genomics is becoming more ubiquitous and will be incorporated into healthcare of individuals along the health spectrum

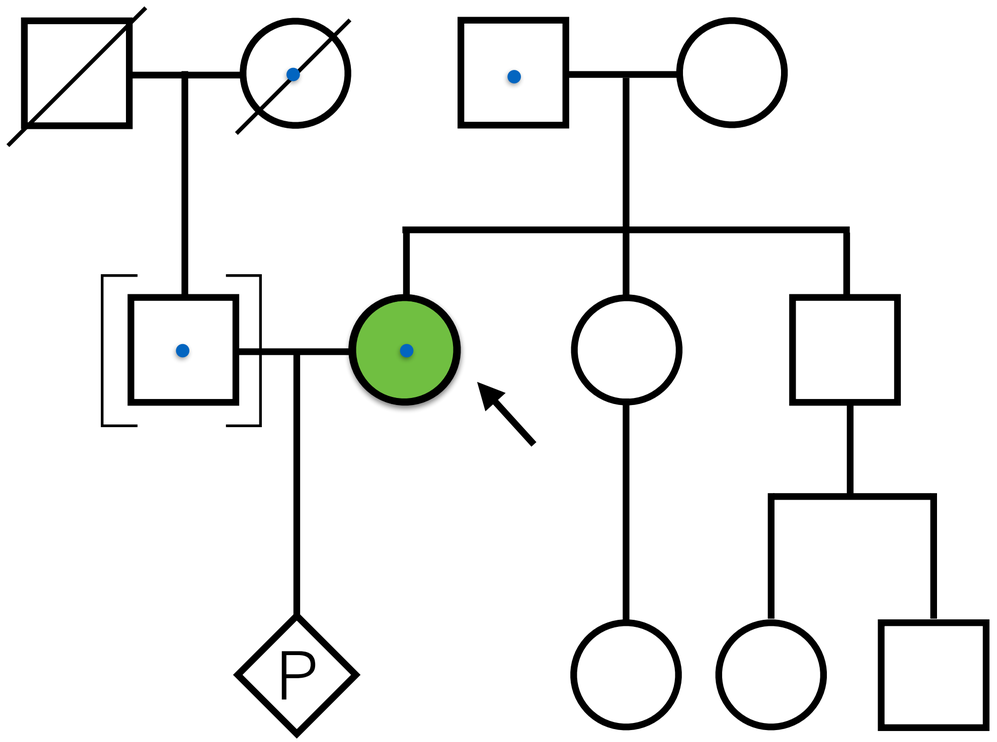
Even if you don't order any genetic tests, a patient will eventually bring in a test report and expect you to know what to do with it



Population: 4.85 million

Common Misconceptions

Misconception 1: “No one in my family has any genetic diseases, so I and my children are not at risk”



Small families

Confounding histories

Unknown family history

De novo pathogenic variants

Recessive conditions

Misconception 2: “I’m the first person in my family with this disease, so it can’t be genetic”

Small families

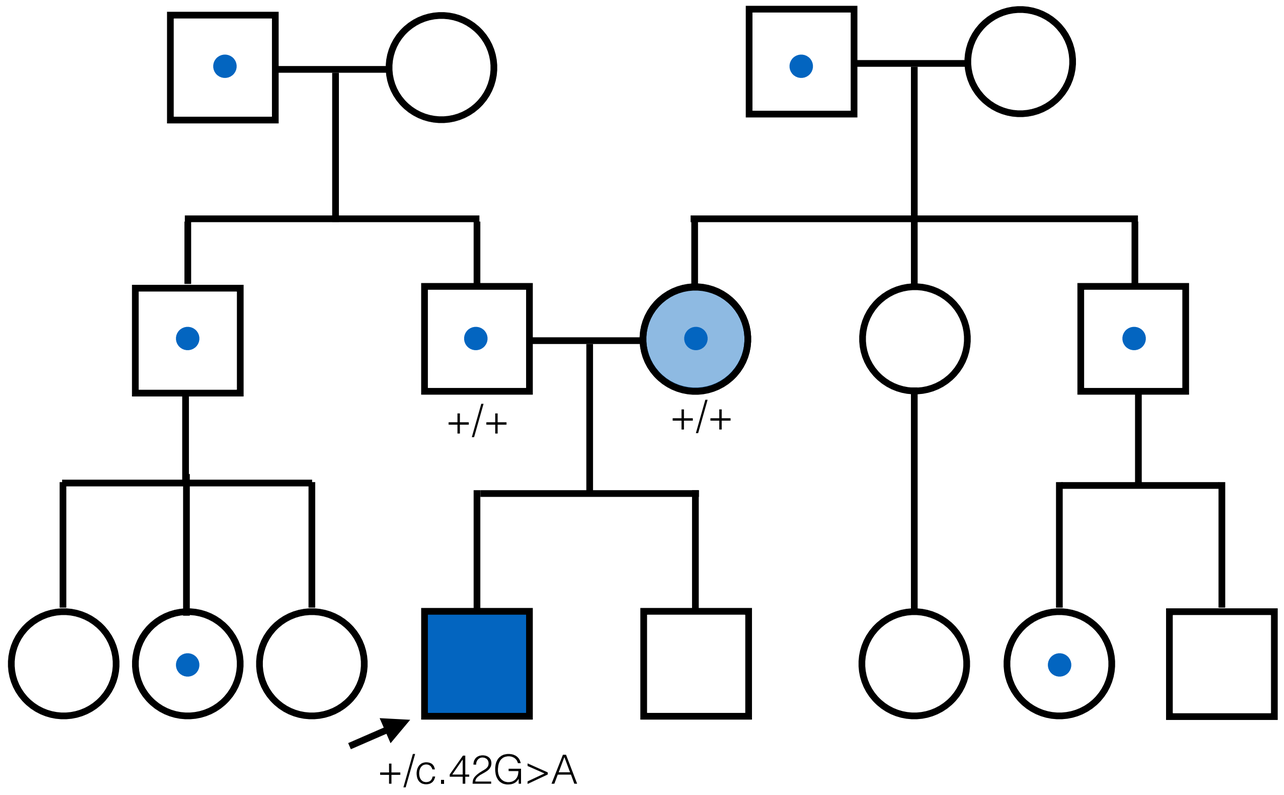
Unknown family history

Reduced penetrance

Variable expressivity

De novo pathogenic variants

Recessive conditions

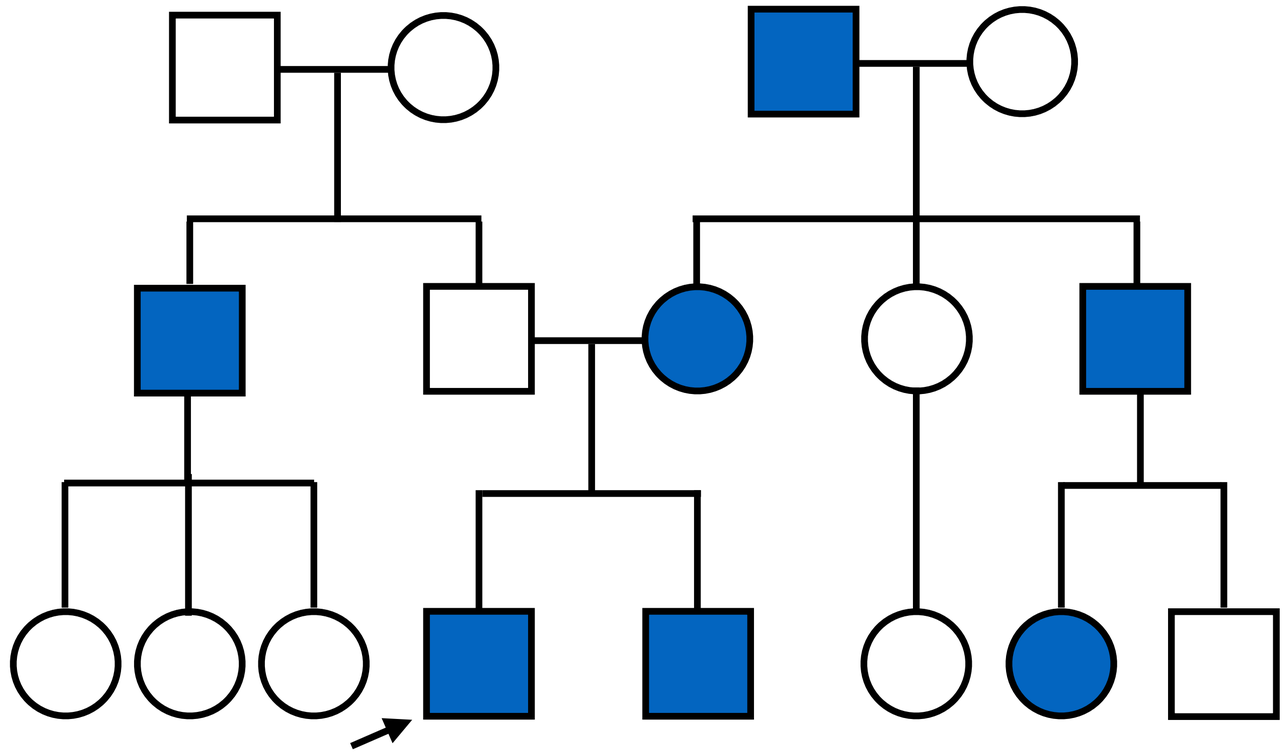


Misconception 3: “This condition runs in my family, so it must be genetic”

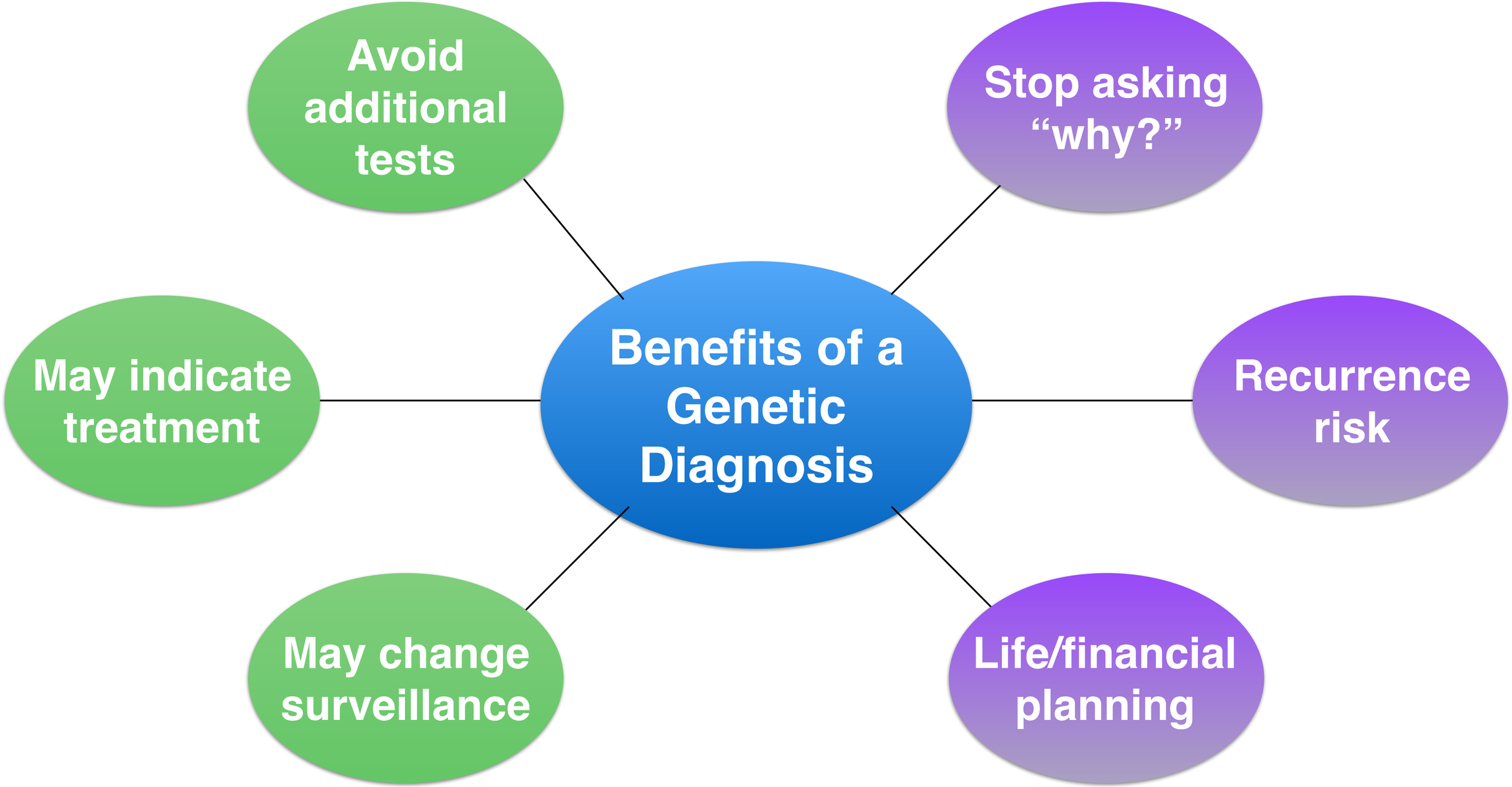
Common diseases

Family members share genes, but also environmental and lifestyle factors

Having affected family members *does* increase risk for many conditions



Misconception 4: “Even if this test is positive, it’s not going to change anything…what’s the point?”

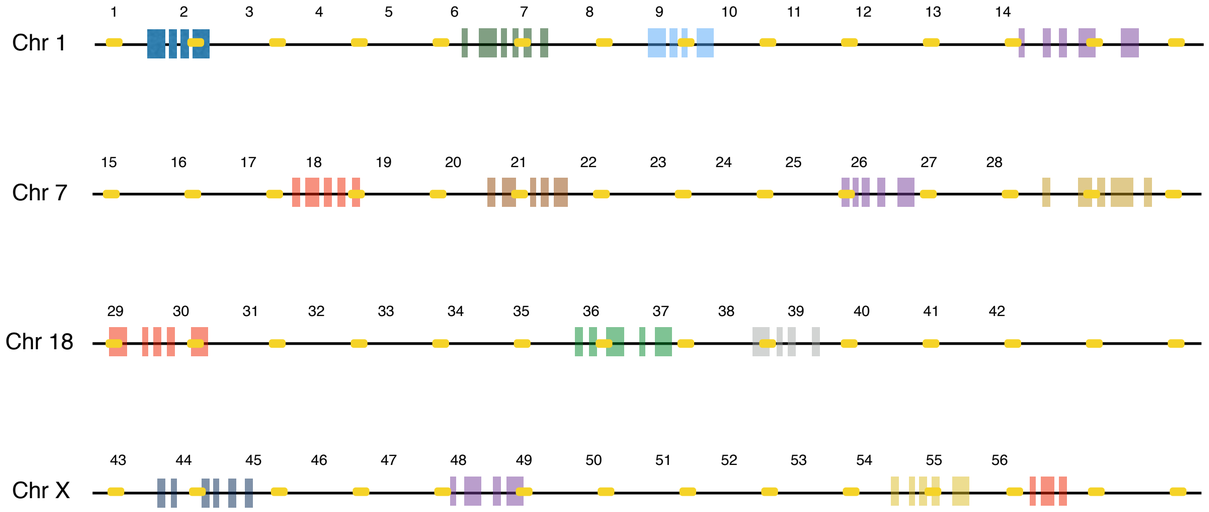

Avoid additional tests
Stop asking "why?"
May indicate treatment
Benefits of a Genetic Diagnosis
Recurrence risk
May change surveillance
Life/financial planning

Misconception 5: “I’ve already had genetic testing, so I don’t need whole genome sequencing”

Karyotype

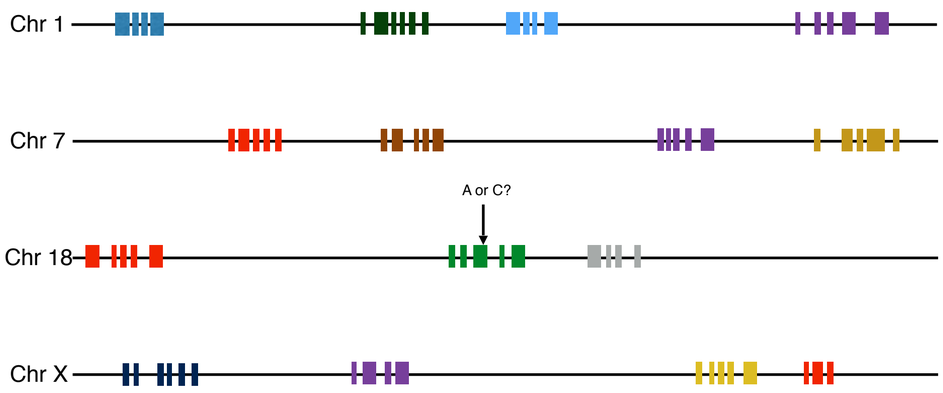
Cost: $500-1200

Microarray

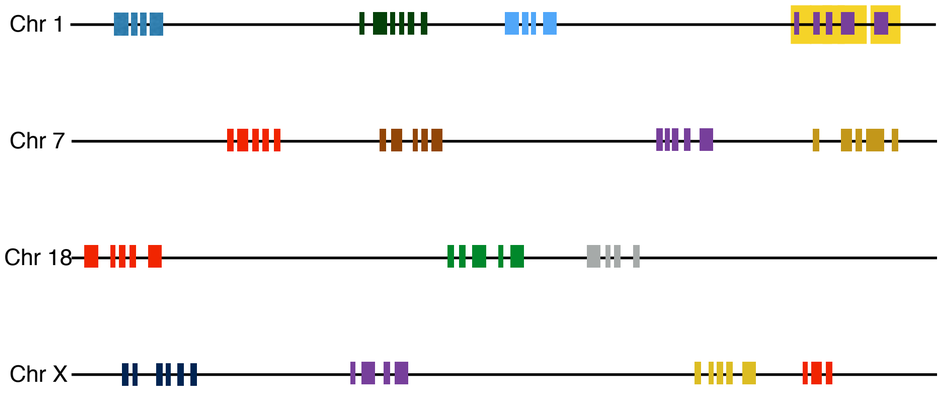


Cost: $1500-3000

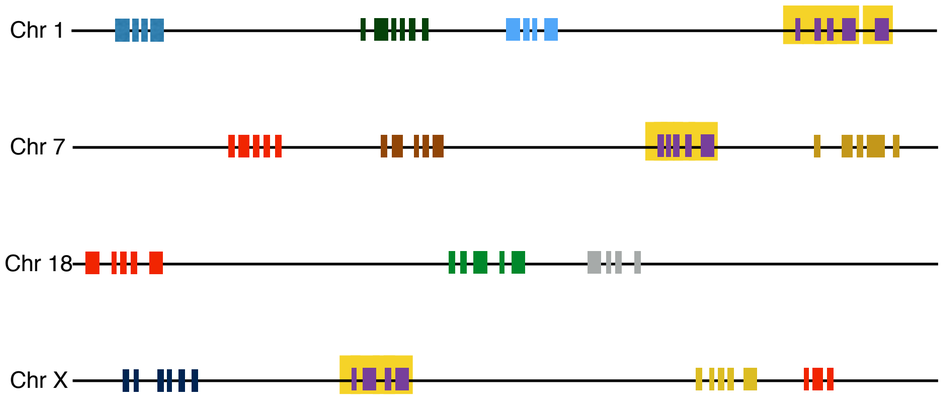
Single site mutation



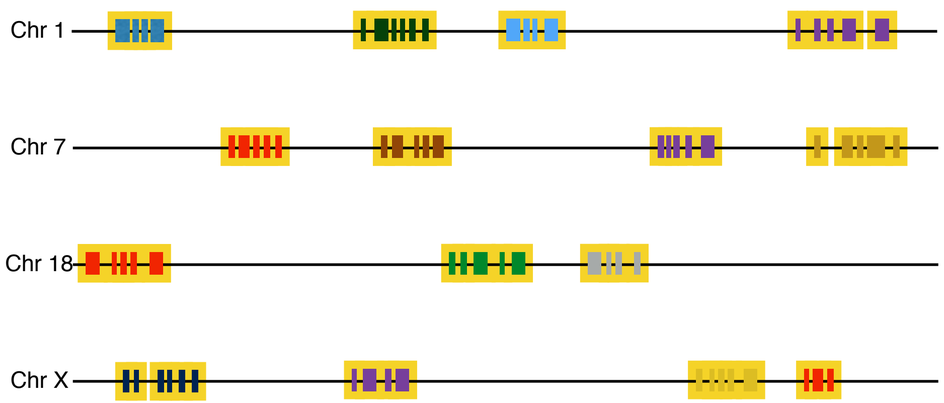
Sequencing a single gene



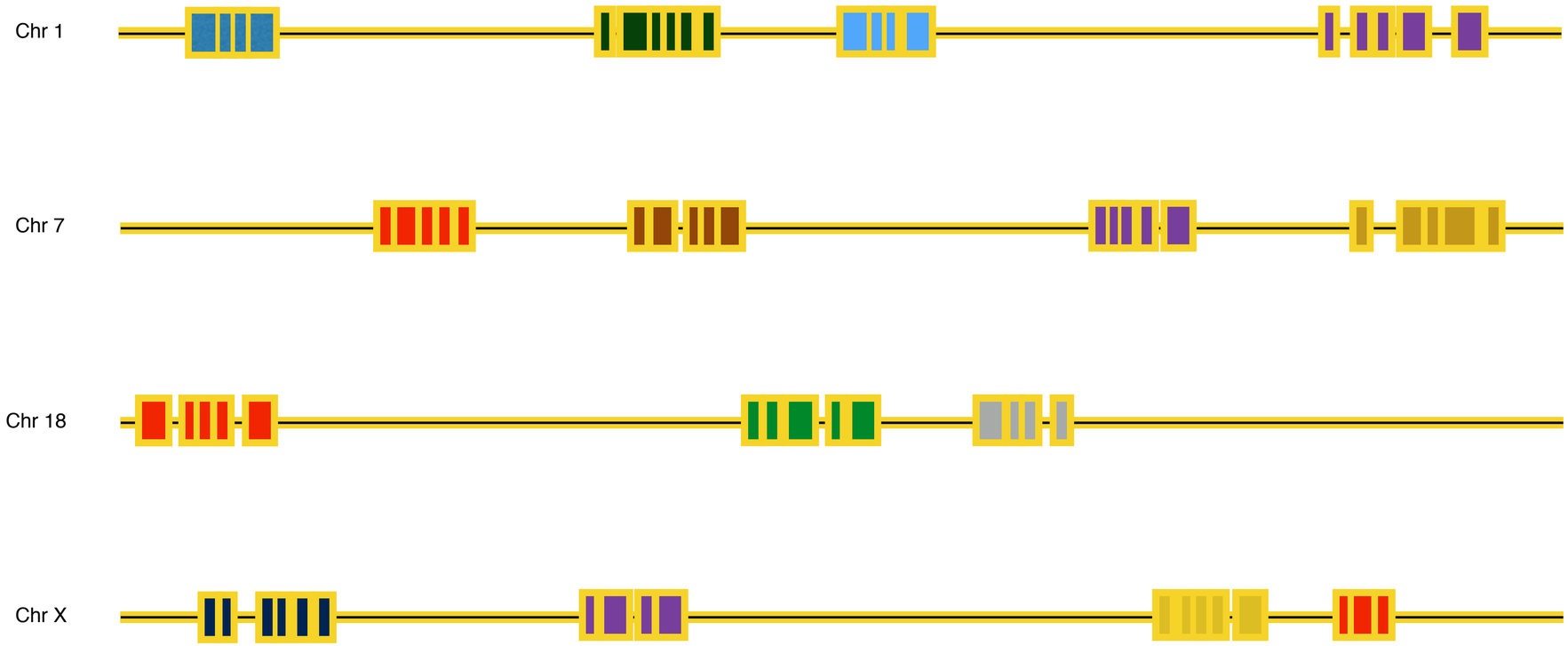
Sequencing a panel of genes



Exome sequencing

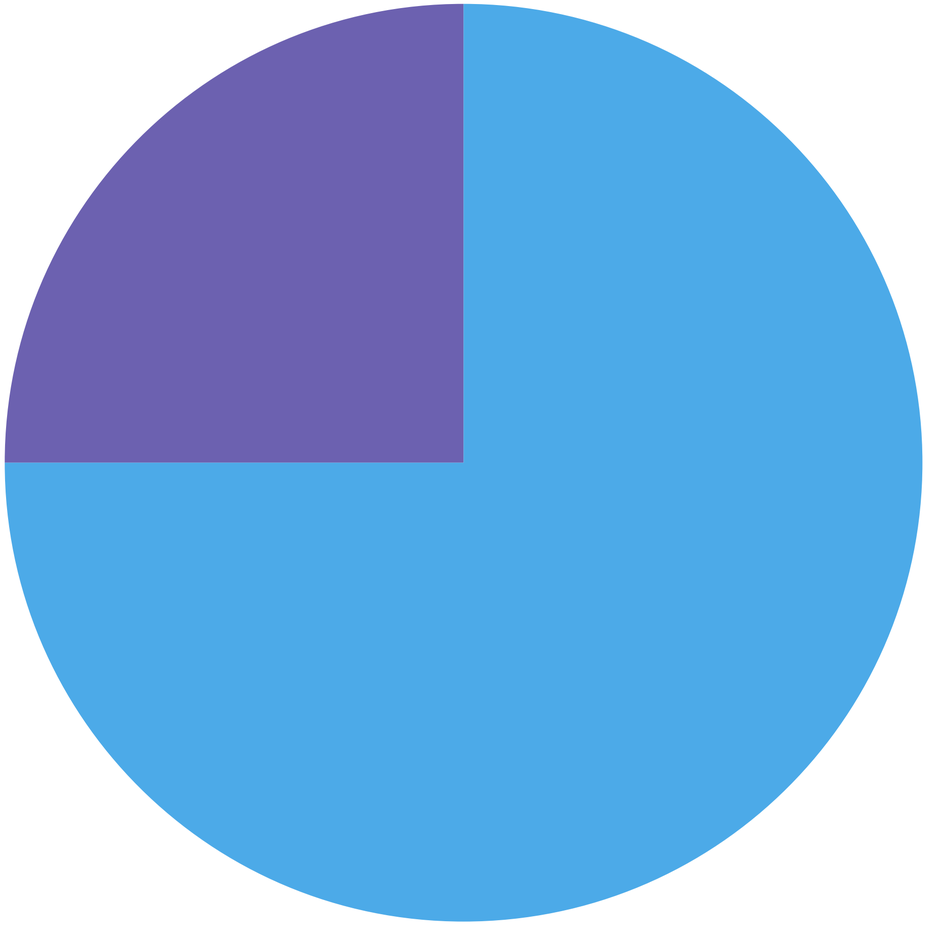


Genome sequencing



Misconception 6: “My whole genome sequencing was negative, so my disease must not be genetic”

Limits of 2016 Knowledge



~20,000 human genes

Only ~5,000 have been definitively associated with human disease

Variants of uncertain significance in genes that are not well known may not be reported

Causative variants may not be identified with WGS due to limitations of technology

Misconception 7: “Whole genome sequencing can tell me about my risk for any/all diseases

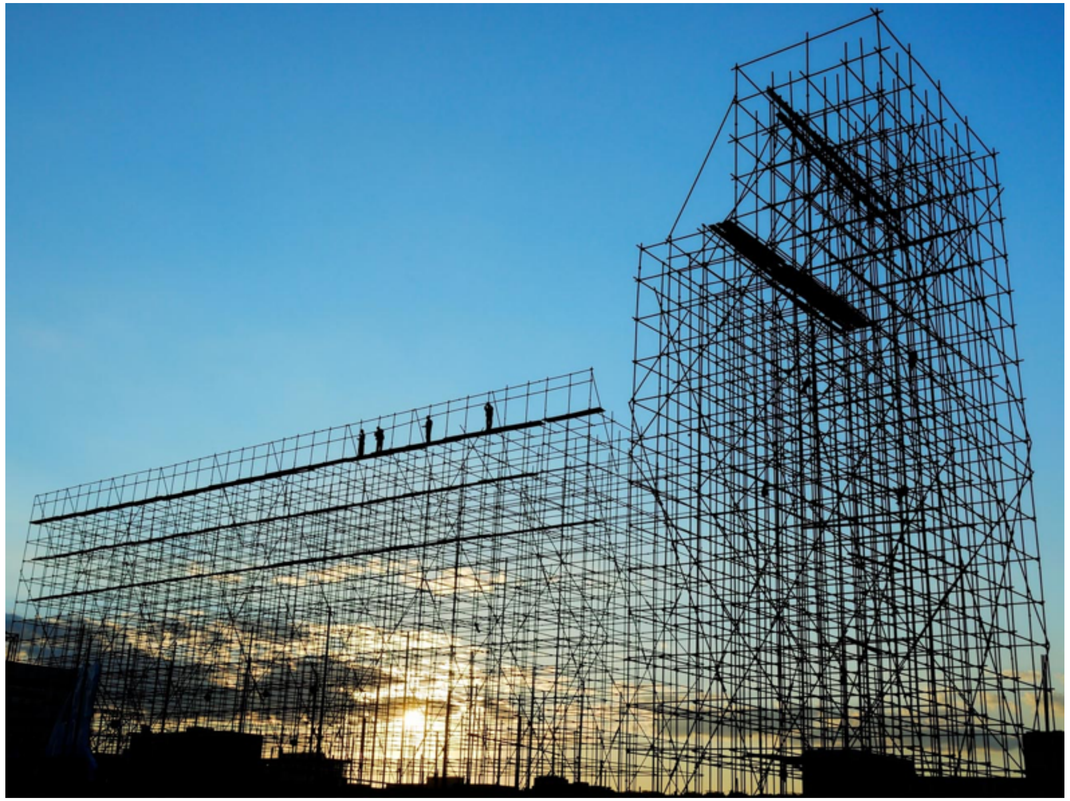
Common, complex diseases

Multifactorial conditions

Threshold model

Dealing with Misconceptions

1. Elicit the misconception
2. Identify the misconception
3. Deconstruct the misconception
4. Replace the misconception



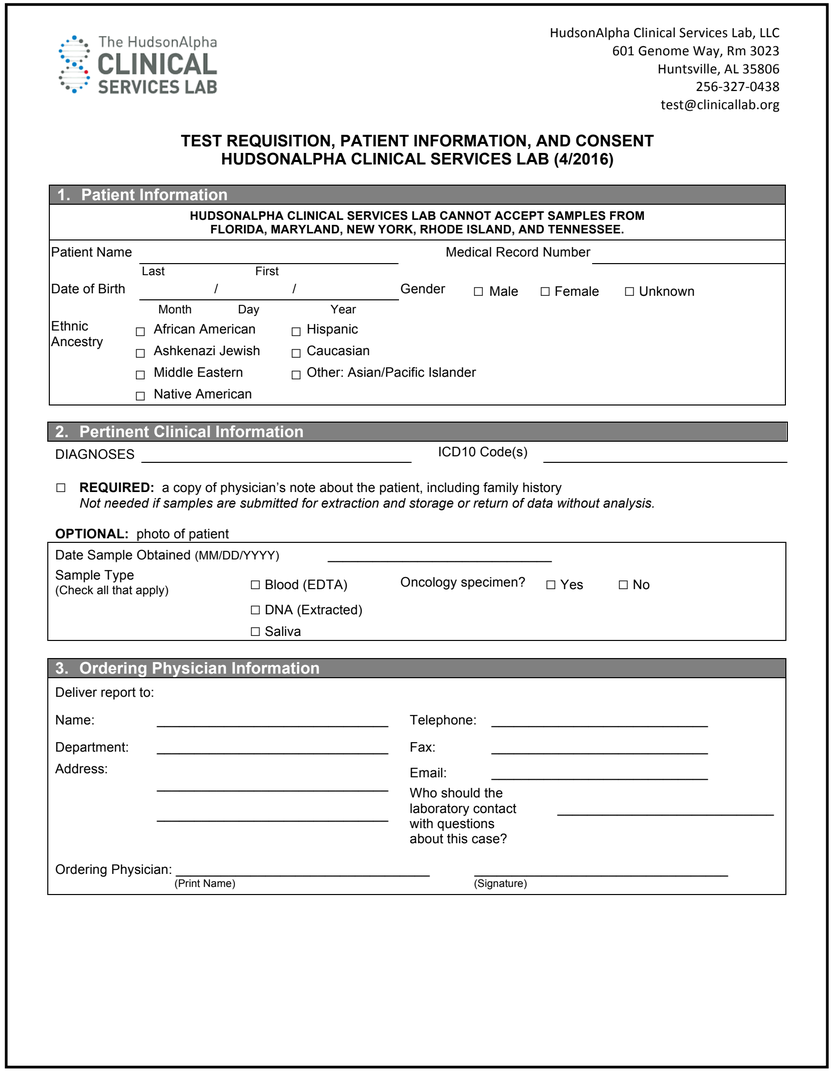
Pre- and Post-Test Discussion Points

Before You Order a Genome:

1. Is there another test that would be more appropriate for your patient?

2. Does your patient have reasonable expectations of the potential results of testing?

3. Has the patient had the opportunity to make appropriate choices regarding testing, and are they confident in their choices?



The HudsonAlpha CLINICAL SERVICES LAB

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601 Genome Way, Rm 3023
Huntsville, AL 35806
256-327-0438
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TEST REQUISITION, PATIENT INFORMATION, AND CONSENT
HUDSONALPHA CLINICAL SERVICES LAB (4/2016)

1. Patient Information

HUDSONALPHA CLINICAL SERVICES LAB CANNOT ACCEPT SAMPLES FROM FLORIDA, MARYLAND, NEW YORK, RHODE ISLAND, AND TENNESSEE.

Patient Name _____ (Last _____ First) Medical Record Number _____

Date of Birth ____ / ____ / ____ (Month Day Year) Gender ☐ Male ☐ Female ☐ Unknown

Ethnic Ancestry
☐ African American ☐ Hispanic
☐ Ashkenazi Jewish ☐ Caucasian
☐ Middle Eastern ☐ Other: Asian/Pacific Islander
☐ Native American

2. Pertinent Clinical Information

DIAGNOSES _____ ICD10 Code(s) _____

☐ **REQUIRED:** a copy of physician's note about the patient, including family history
Not needed if samples are submitted for extraction and storage or return of data without analysis.

OPTIONAL: photo of patient

Date Sample Obtained (MM/DD/YYYY) _____

Sample Type (Check all that apply)
☐ Blood (EDTA)
☐ DNA (Extracted)
☐ Saliva

Oncology specimen? ☐ Yes ☐ No

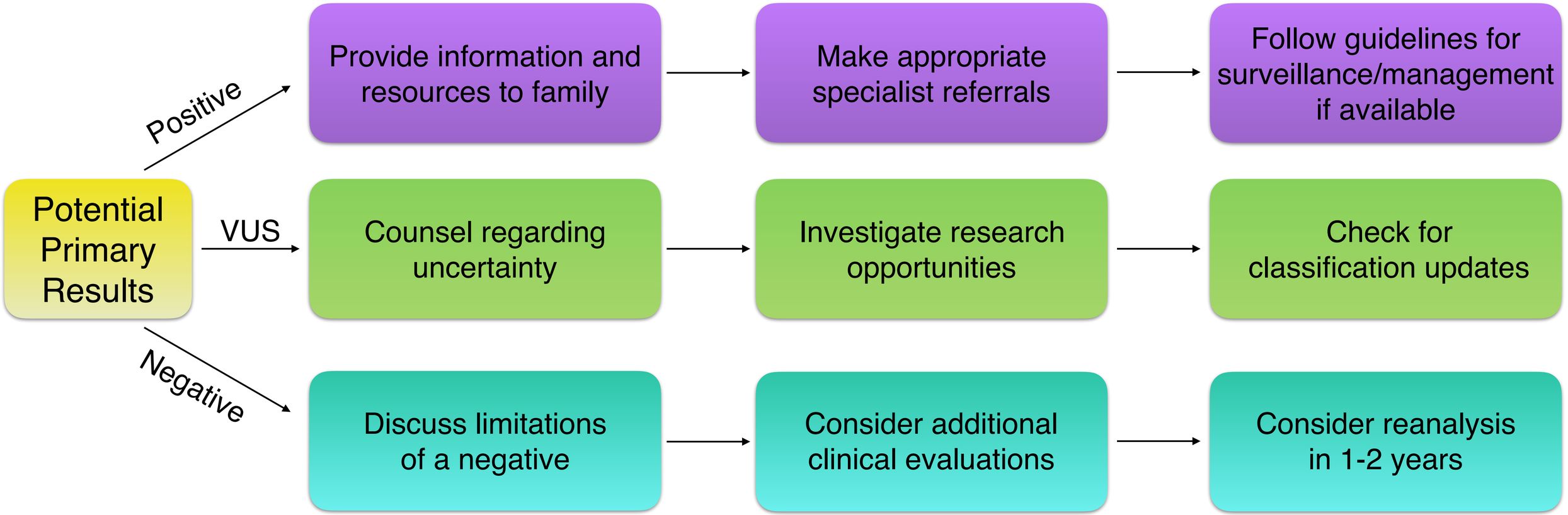
3. Ordering Physician Information

Deliver report to:

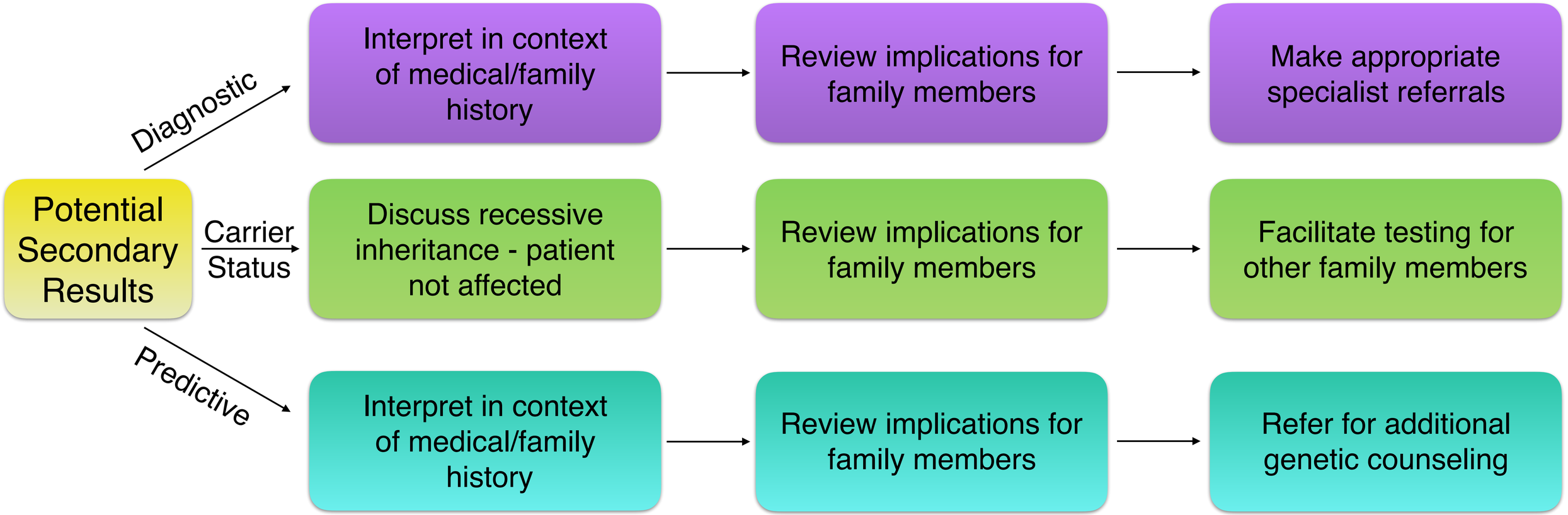
Name: _____ Telephone: _____
Department: _____ Fax: _____
Address: _____ Email: _____
_____ Who should the laboratory contact with questions about this case? _____
_____

Ordering Physician: _____ (Print Name) _____ (Signature)

After You Receive a Report - Primary Findings



After You Receive a Report - Secondary Findings



Where Can I Get More Information?

SIMONS VIP CONNECT

Genetics Home Reference

NIH National Human Genome Research Institute

The Smith Family CLINIC for GENOMIC MEDICINE

P: 256-327-9640
Web: smithfamilyclinic.org

The HudsonAlpha CLINICAL SERVICES LAB

P: 256-327-9670
Web: clinicallab.org

Questions?