

Genomics in Primary Care: The Promise of Personalized Medicine

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THE PROMISE OF PERSONALIZED MEDICINE

FMEC CONFERENCE · NOVEMBER 1ST 2013
PHILADELPHIA, PA

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BACKGROUND

- Whole genome sequencing
 - Human Genome Project completed in 2003
- Personalized Medicine
 - Predictive Medicine (Risk)
 - Pharmacogenomics
- Move from single gene to multiple gene
 - Understanding of disease

BACKGROUND

- Direct to Consumer testing
- <https://www.23andme.com/health/all/>



Carrier Status (53)

ARSACS *

Agnesis of the Corpus Callosum with Peripheral Neuropathy (ACCPN) *

Alpha-1 Antitrypsin Deficiency *

Autosomal Recessive Polycystic Kidney Disease *

BRCA Cancer Mutations (Selected) *

Beta Thalassemia *

Bloom's Syndrome *

Canavan Disease *

Congenital Disorder of Glycosylation Type 1a (PMM2-CDG) *

Connexin 26-Related Sensorineural Hearing Loss *

Cystic Fibrosis *

D-Bifunctional Protein Deficiency *

DPD Deficiency *

Dihydropyrimidinase Dehydrogenase Deficiency *

Factor XI Deficiency *

Familial Dysautonomia *

Familial Hypercholesterolemia Type B *

Familial Hyperinsulinism (ABCC8-related) *

Familial Mediterranean Fever *

Fanconi Anemia (FANCC-related) *

G6PD Deficiency *

GRACILE Syndrome *

Gaucher Disease *

Glycogen Storage Disease Type 1a *

Glycogen Storage Disease Type 1b *

Hemochromatosis (HFE-related) *

Hereditary Fructose Intolerance *

Hypertrophic Cardiomyopathy (MYBPC3 25bp-deletion) *

LAMB3-related Junctional Epidermolysis Bullosa *

Leigh Syndrome, French Canadian Type (LSFC) *

Limb-girdle Muscular Dystrophy *

Maple Syrup Urine Disease Type 1B *

Medium-Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency *

Mucopolipidosis IV *

Neuronal Ceroid Lipofuscinosis (CLN5-related) *

Neuronal Ceroid Lipofuscinosis (PPT1-related) *

Niemann-Pick Disease Type A *

Nijmegen Breakage Syndrome *

Pendred Syndrome *

Phenylketonuria *

Primary Hyperoxaluria Type 2 (PH2) *

Rhizomelic Chondrodysplasia Punctata Type 1 (RCDP1) *

Salla Disease *

Sickle Cell Anemia & Malaria Resistance *

Drug Response (24)

Abacavir Hypersensitivity *

Alcohol Consumption, Smoking and Risk of Esophageal Cancer *

Antidepressant Response

Beta-Blocker Response

Caffeine Metabolism

Clopidogrel (Plavix®) Efficacy *

Floxacin Toxicity

Fluorouracil Toxicity *

Hepatitis C Treatment Side Effects

Heroin Addiction

Lumiracoxib (Prexige®) Side Effects

Metformin Response

Naltrexone Treatment Response

Oral Contraceptives, Hormone Replacement Therapy and Risk of Venous Thromboembolism *

Phenytoin (Dilantin®) Sensitivity (Epilepsy Drug) *

Postoperative Nausea and Vomiting (PONV)

Pseudocholinesterase Deficiency *

Response to Hepatitis C Treatment *

Response to Interferon Beta Therapy

Statin Response

Sulfonylurea Drug Clearance (Type 2 Diabetes Treatment) *

Thiopurine Methyltransferase Deficiency *

Warfarin (Coumadin®) Sensitivity *

Warfarin (Coumadin®) Sensitivity: Preliminary Research

Traits (60)

Adiponectin Levels

Alcohol Flush Reaction *

Asparagus Metabolite Detection

Avoidance of Errors

Biological Aging

Birth Weight

Bitter Taste Perception *

Blood Glucose

Breast Morphology

Breastfeeding and IQ

C-reactive Protein Level

Caffeine Consumption

Childhood and Adolescent Growth

Chronic Hepatitis B

Earwax Type *

Eating Behavior

Eye Color *

Eye Color: Preliminary Research

Finger Length Ratio

Food Preference

Freckling

HDL ("Good") Cholesterol Levels

HIV Progression

Hair Color

Hair Curl *

Hair Curl: Preliminary Research

Hair Thickness

Height

Hypospadias

Iris Patterns

LDL ("Bad") Cholesterol Levels

Lactose Intolerance *

Leprosy Susceptibility

Longevity

Malaria Complications

Malaria Resistance (Duffy Antigen) *

Male Pattern Baldness *

Male Pattern Baldness: Preliminary Research

Measures of Intelligence

Measures of Obesity

Memory

Menarche

Menopause

Muscle Performance *

Disease Risk (122)

Abdominal Aortic Aneurysm

Age-related Macular Degeneration *

Alcohol Dependence

Alopecia Areata

Alzheimer's Disease *

Alzheimer's Disease: Preliminary Research

Ankylosing Spondylitis

Asthma

Atopic Dermatitis

Atrial Fibrillation *

Atrial Fibrillation: Preliminary Research

Attention-Deficit Hyperactivity Disorder

Back Pain

Basal Cell Carcinoma

Behçet's Disease

Bipolar Disorder *

Bipolar Disorder: Preliminary Research

Bladder Cancer

Brain Aneurysm

Breast Cancer *

Breast Cancer Risk Modifiers

Celiac Disease *

Celiac Disease: Preliminary Research

Chronic Kidney Disease *

Chronic Lymphocytic Leukemia

Chronic Obstructive Pulmonary Disease (COPD)

Cleft Lip and Cleft Palate

Cluster Headaches

Colorectal Cancer *

Coronary Heart Disease *

Coronary Heart Disease: Preliminary Research

Creutzfeldt-Jakob Disease

Crohn's Disease *

Developmental Dyslexia

Dupuytren's Disease

Endometriosis

Esophageal Cancer: Preliminary Research

Esophageal Squamous Cell Carcinoma (ESCC) *

Essential Tremor

Exfoliation Glaucoma *

Follicular Lymphoma

Gallstones *

Generalized Vitiligo

Gestational Diabetes

Glaucoma: Preliminary Research

Gout *

Gout: Preliminary Research

Hashimoto's Thyroiditis

Hay Fever (Allergic Rhinitis)

Heart Rhythm Disorders (Arrhythmias)

High Blood Pressure (Hypertension)

Hodgkin Lymphoma

Hypertriglyceridemia

Hypothyroidism

Intrahepatic Cholestasis of Pregnancy

Keloid

Kidney Cancer

Kidney Disease

Kidney Stones

Larynx Cancer

Lou Gehrig's Disease (ALS)

Lung Cancer *

Lupus (Systemic Lupus Erythematosus) *

Male Breast Cancer

Male Infertility

Melanoma *

Melanoma: Preliminary Research

Meningioma

Migraines

Multiple Sclerosis *

Myeloproliferative Neoplasms

Narcolepsy

Nasopharyngeal Carcinoma

Neural Tube Defects

Neuroblastoma

Nicotine Dependence

Nonalcoholic Fatty Liver Disease

Obesity *

Obesity: Preliminary Research

Obsessive-Compulsive Disorder

Oral and Throat Cancer

Osteoarthritis

Otosclerosis

Ovarian Cancer

Paget's Disease of Bone

Pancreatic cancer

Parkinson's Disease *

Parkinson's Disease: Preliminary Research

Peripheral Arterial Disease

Placental Abruption

Polycystic Ovary Syndrome

Preeclampsia

Primary Biliary Cirrhosis *

Primary Biliary Cirrhosis: Preliminary Research

Progressive Supranuclear Palsy

Prostate Cancer *

Psoriasis *

Pulmonary Fibrosis

Restless Legs Syndrome *

Restless Legs Syndrome: Preliminary Research

Rheumatoid Arthritis *

Sarcoidosis

Sarcoma

Schizophrenia

Scleroderma (Limited Cutaneous Type) *

Scoliosis

Selective IgA Deficiency

Sjögren's Syndrome

Squamous Cell Carcinoma

Stomach Cancer (Gastric Cardia Adenocarcinoma) *

Stomach Cancer: Preliminary Research

Stroke

Sudden Cardiac Arrest

Tardive Dyskinesia

Testicular Cancer

Thyroid Cancer

Tourette's Syndrome

Type 1 Diabetes *

Type 2 Diabetes *

Ulcerative Colitis *

Uterine Fibroids

Venous Thromboembolism *

METHODS

- Mailed paper survey to primary care physicians
- 3 regional health systems
 - Christiana Care Health System plus DAFP
 - Jefferson Health System
 - Lehigh Valley Health Network
- Introductory letter with all 3 logos
- \$5 Wawa gift card
- Supported by Dean's office at TJU

RESULTS

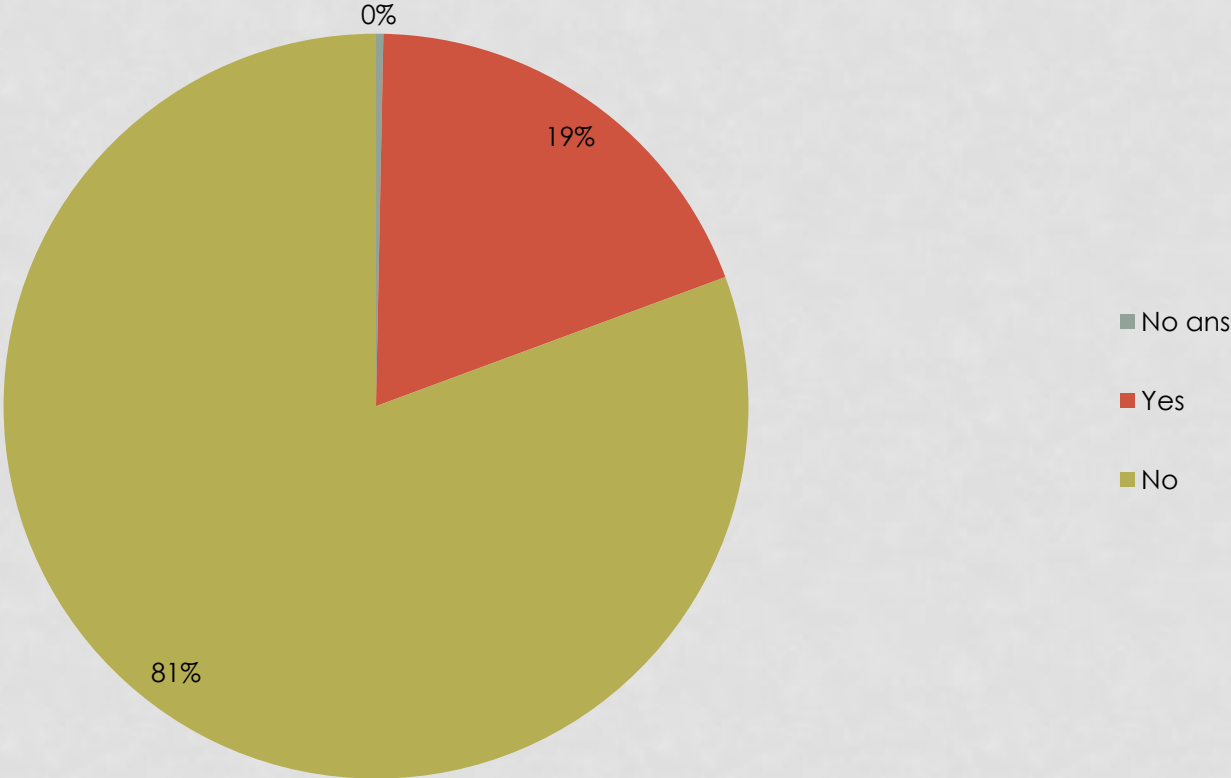
- $n = 316$
- Response rate 37.89%
- 67% Academic
- 33% Private

PARTICIPANTS' DESCRIPTION

(n= 316)

<u>Male</u>	53%
<u>Age</u>	
25-34	16%
35-44	25%
45-54	28%
55-64	26%
65-74	4%
≥ 75	2%
<u>Specialty</u>	
FM	67%
IM	23%
Other	10%

IN THE LAST 6 MONTHS HAVE YOU ORDERED GENETIC TESTING?



GENETIC TESTING ORDERED

Testing	(n)
Cancer Risk	28
Prenatal	11
Other*	11
Pharmacogenomics	9
Micro array	6

PHYSICIAN CONFIDENCE

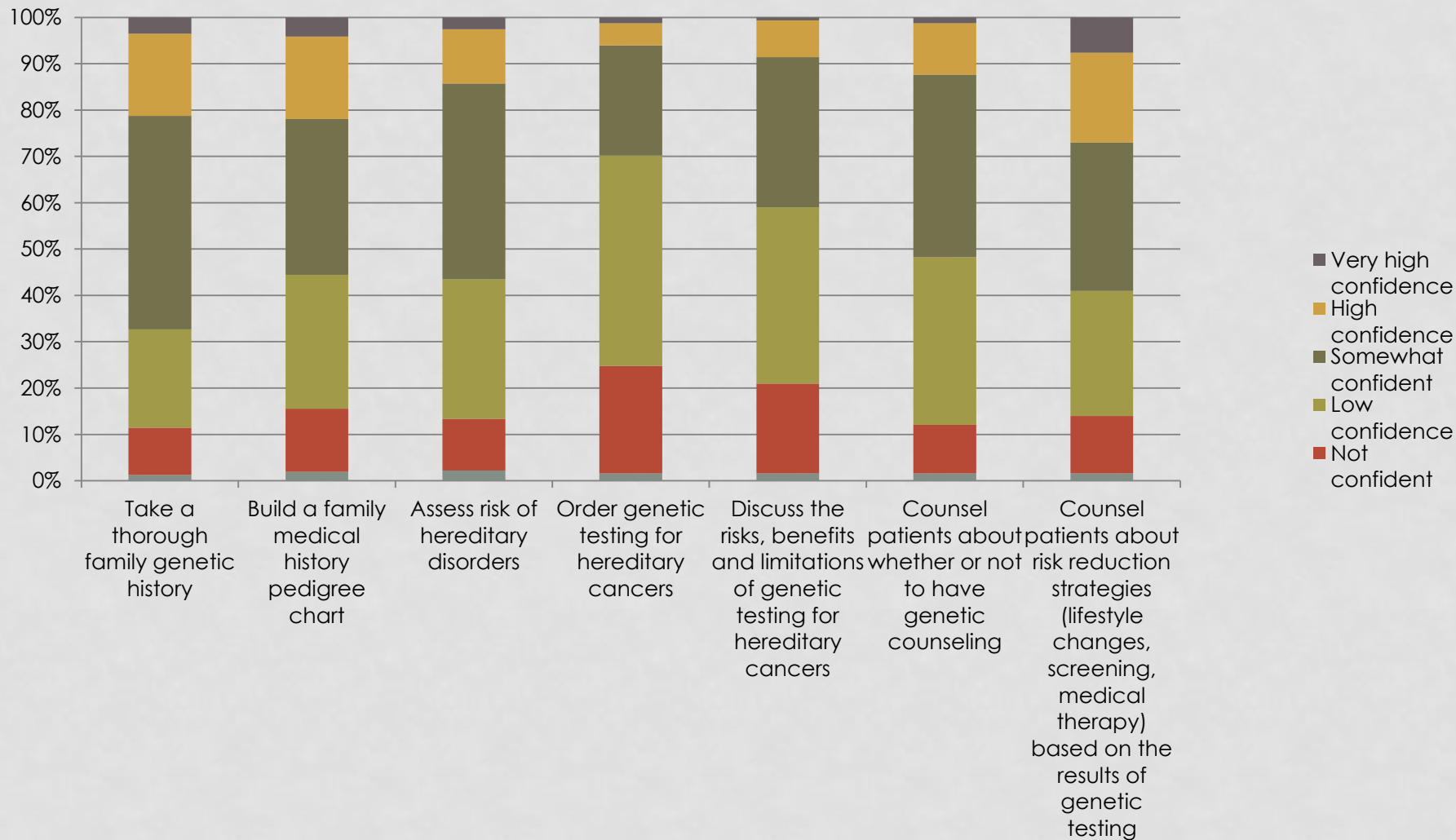


TABLE A

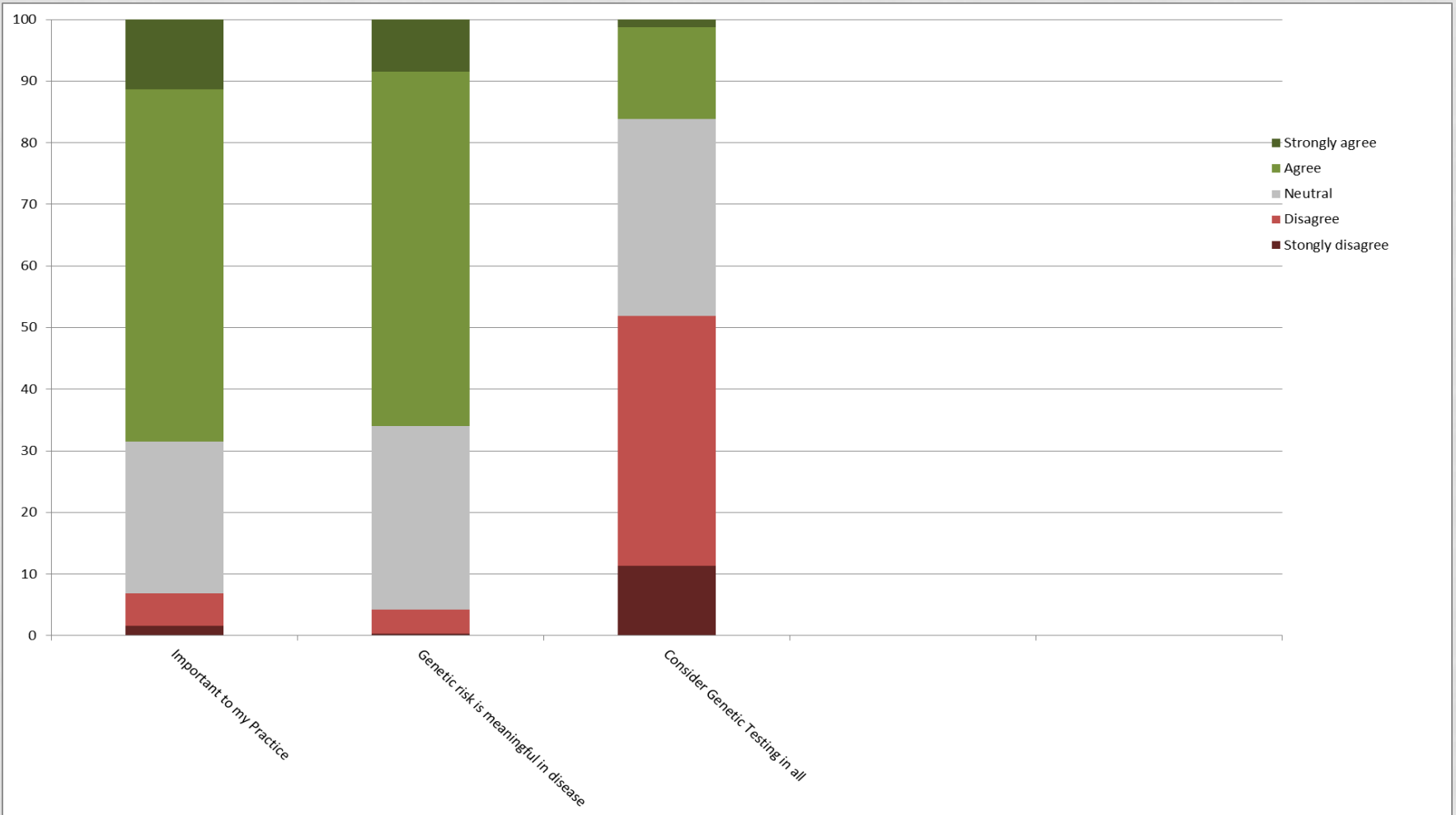
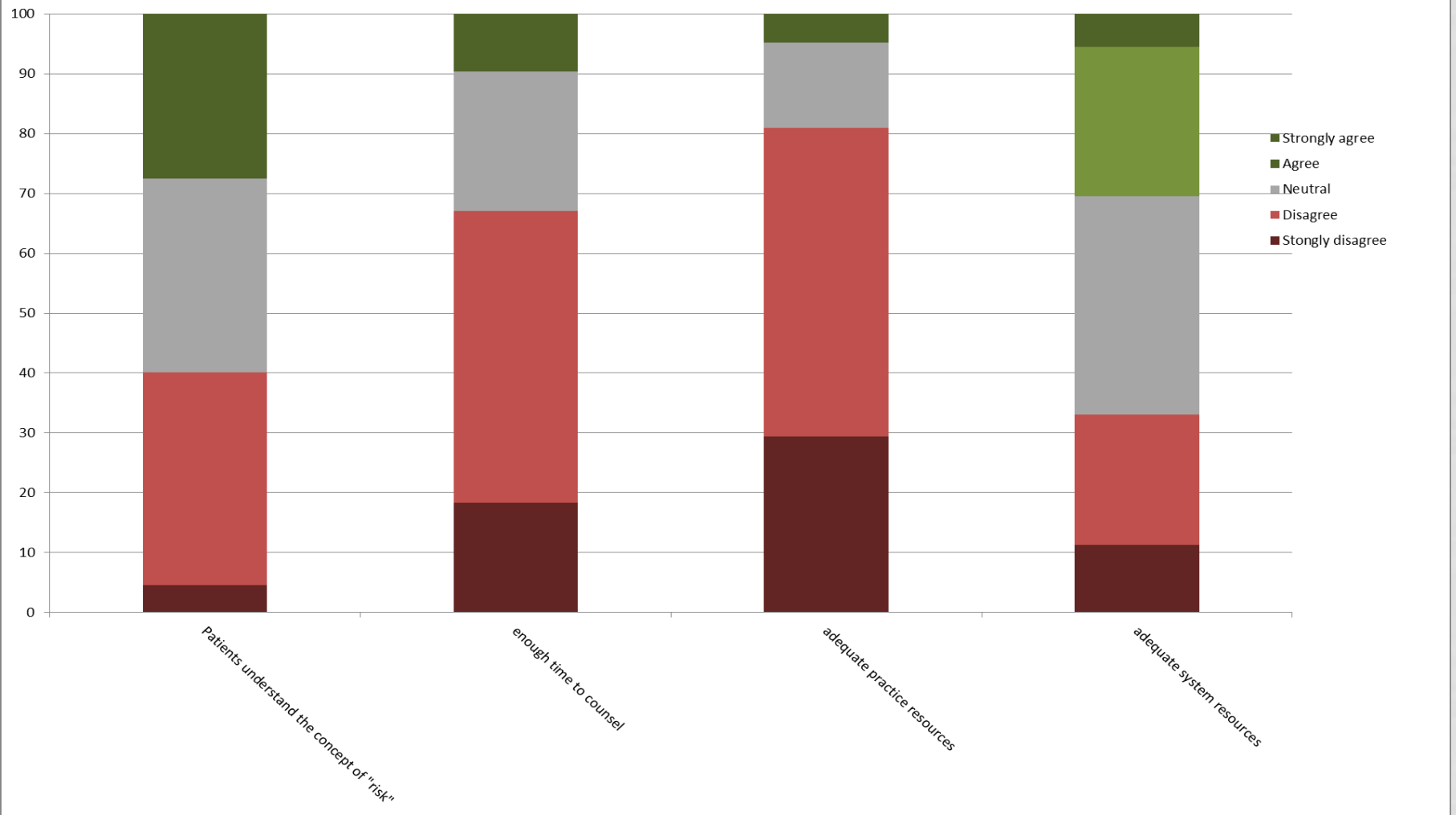


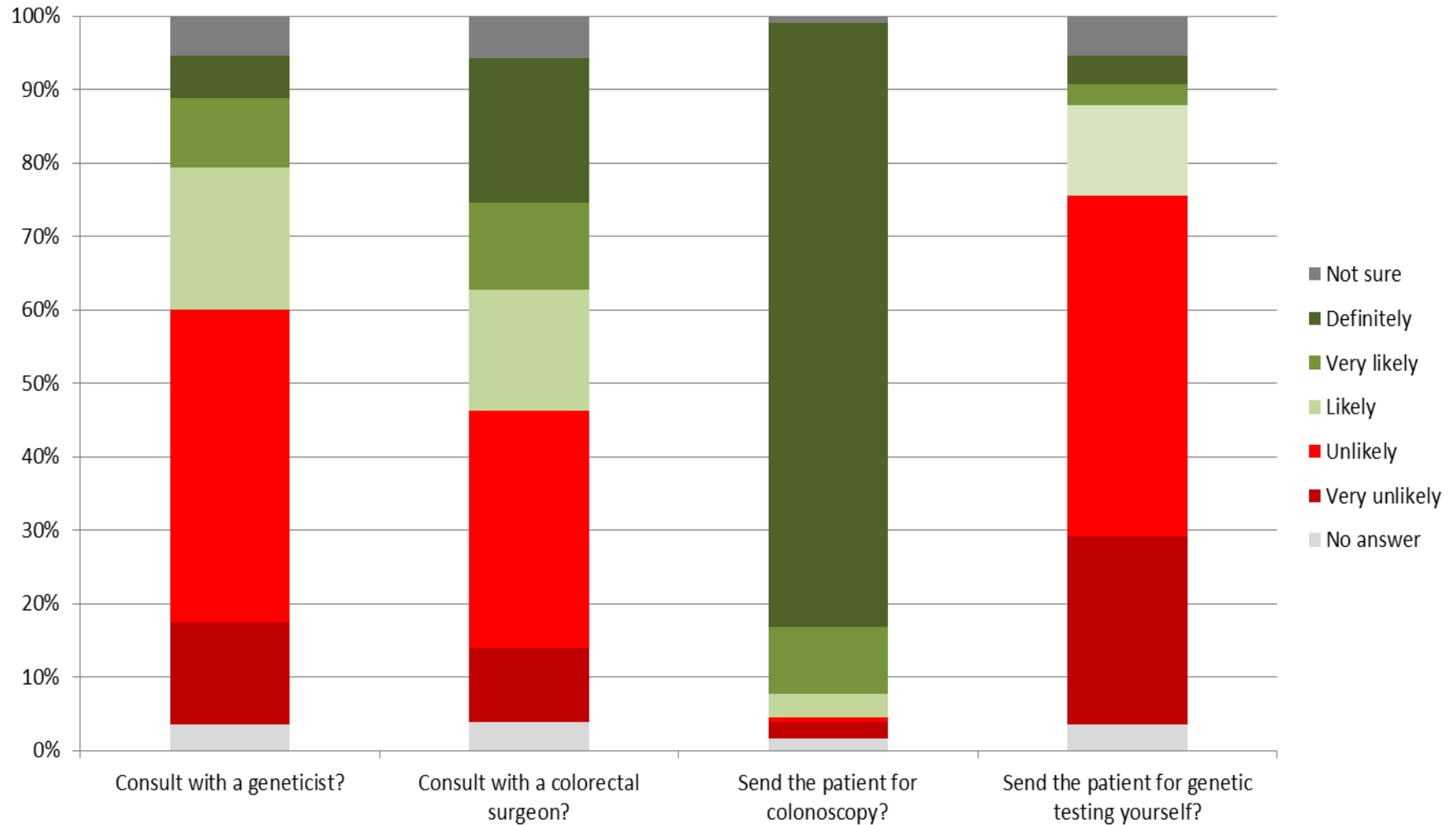
TABLE B



CASE 1

A 43 year-old woman comes to you concerned about her risk for cancer. You obtain a family history and discover that her father had colorectal cancer diagnosed at age 49 and a sister diagnosed with colorectal cancer at 45 years of age.

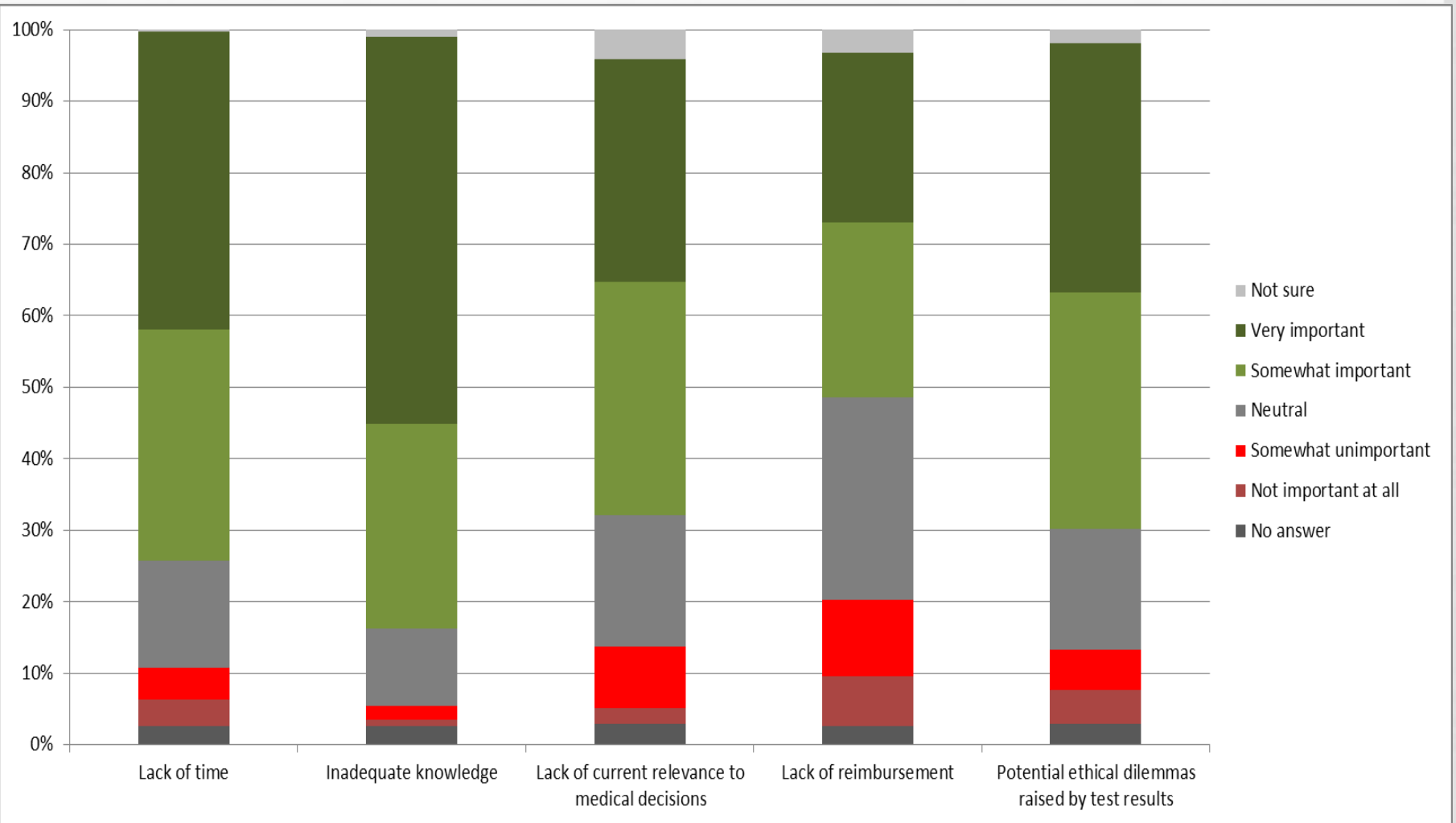
HOW LIKELY WOULD YOU BE TO...



CASE 2

- A 52 year old healthy man with an unremarkable family history tells you that he has had his blood drawn for a “full genetic analysis” (currently commercially available for less than \$5000.) He wants to bring the results in to talk to you about your recommendations.

HOW IMPORTANT ARE THE FOLLOWING BARRIERS TO DISCUSSING HIS RESULTS?

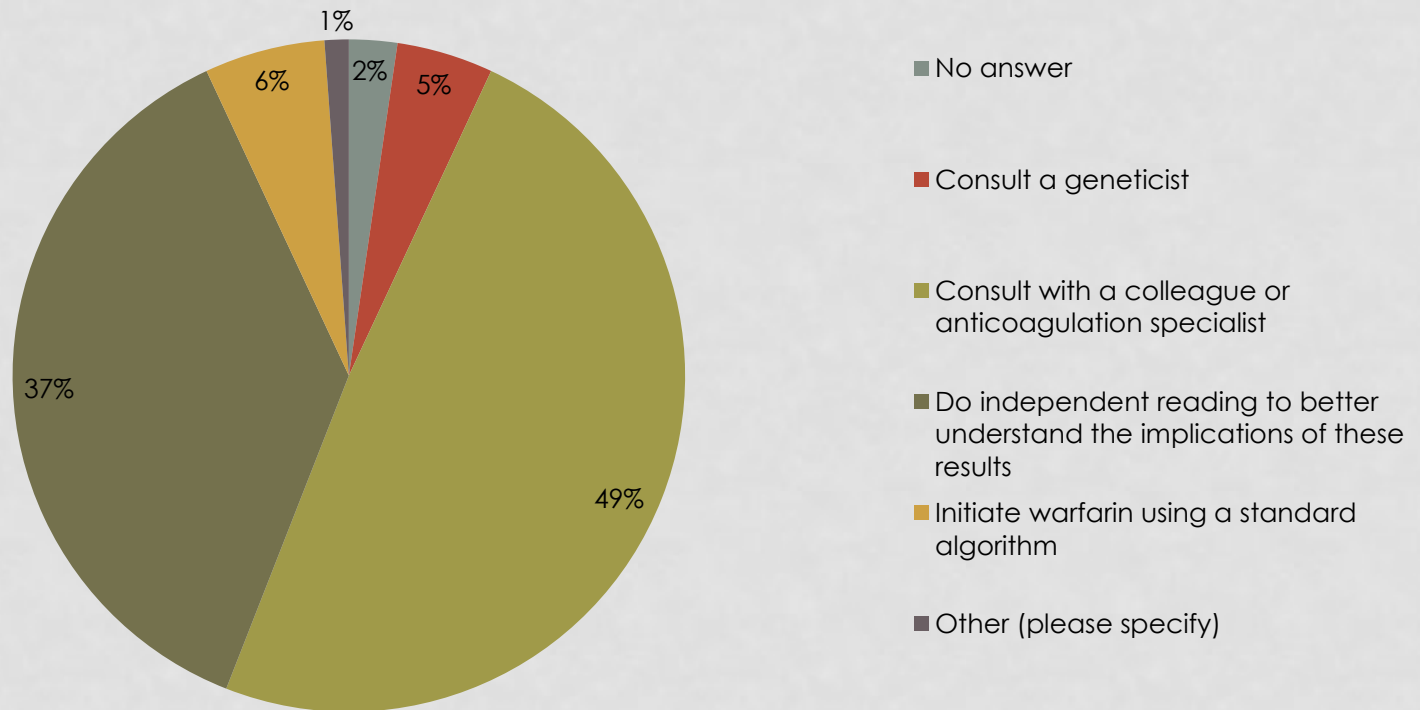


CASE 3

- You are seeing a 57-year old woman with a recent DVT for warfarin management. A lab report from her recent hospitalization includes information about genomic markers relevant to warfarin metabolism.

YOU ARE MOST LIKELY GOING TO:

Case 3



NEXT STEPS

- Current state of Family History data in Electronic Medical Record
- Availability and integration of resources at the point of care
- Challenge of education initiatives in real world primary care

USEFUL LINKS

- GeneTests (select “GeneReviews”) : <http://www.genetests.org>
- OMIM (Online Mendelian Inheritance in Man) :
<http://www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM>
- Genes and Disease (NCBI) (organized by parts of the body affected):
<http://www.ncbi.nlm.nih.gov/books/bv.fcgi?rid=gnd>
- Chromosomal Variation in Man:
<http://www.wiley.com/legacy/products/subject/life/borgaonkar/access.html>
- National Coalition for Health Professional Education in Genetics:
<http://www.nchpeg.org>
- GeneTests (select “Educational Materials” and “Genetic Tools”):
<http://www.genetests.org>
- March of Dimes—Genetics and Your Practice:
<http://marchofdimes.com/gyponline/index.bm2>

THANK YOU