Genomics in Primary Care: The Promise of Personalized Medicine

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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
Direct to Consumer testing

https://www.23andme.com/health/all/
Carrier Status (53)

ARSACS *
Agenesis 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 *
Dihydrolipoamide 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 *
Mucolipidosis 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 *
Fluorouracil 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)
Pseudocholesterase 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 *
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)

<table>
<thead>
<tr>
<th>Male</th>
<th>53%</th>
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<tbody>
<tr>
<td>Age</td>
<td></td>
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<tr>
<td>25-34</td>
<td>16%</td>
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<tr>
<td>35-44</td>
<td>25%</td>
</tr>
<tr>
<td>45-54</td>
<td>28%</td>
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<tr>
<td>55-64</td>
<td>26%</td>
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<tr>
<td>65-74</td>
<td>4%</td>
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<tr>
<td>≥ 75</td>
<td>2%</td>
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<tr>
<td>Specialty</td>
<td></td>
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<tr>
<td>FM</td>
<td>67%</td>
</tr>
<tr>
<td>IM</td>
<td>23%</td>
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<tr>
<td>Other</td>
<td>10%</td>
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IN THE LAST 6 MONTHS HAVE YOU ORDERED GENETIC TESTING?

- No ans: 81%
- Yes: 19%
- No: 0%
<table>
<thead>
<tr>
<th>Testing</th>
<th>(n)</th>
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</thead>
<tbody>
<tr>
<td>Cancer Risk</td>
<td>28</td>
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<tr>
<td>Prenatal</td>
<td>11</td>
</tr>
<tr>
<td>Other*</td>
<td>11</td>
</tr>
<tr>
<td>Pharmacogenomics</td>
<td>9</td>
</tr>
<tr>
<td>Micro array</td>
<td>6</td>
</tr>
</tbody>
</table>
Take a thorough family genetic history
Build a family medical history pedigree chart
Assess risk of hereditary disorders
Order genetic testing for hereditary cancers
Discuss the risks, benefits and limitations of genetic testing for hereditary cancers
Counsel patients about whether or not to have genetic counseling
Counsel patients about risk reduction strategies (lifestyle changes, screening, medical therapy) based on the results of genetic testing
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.
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

- 49%: Initiate warfarin using a standard algorithm
- 37%: Do independent reading to better understand the implications of these results
- 6%: Consult a geneticist
- 2%: Consult with a colleague or anticoagulation specialist
- 5%: Other (please specify)
- 1%: No answer
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

- National Coalition for Health Professional Education in Genetics: [http://www.nchpeg.org](http://www.nchpeg.org)
THANK YOU