Finding Just Right: Balancing Provider and Payer Goals for Hereditary Genetic Testing
Topics

- Define the needs and goals of genetic testing
  - Payer
  - Provider
- Challenges/limitations
  - Payer
  - Provider
- Insurance coverage and genetic testing
- Future directions
Bridging the gaps

- Healthcare systems
- Evidence-based care
- How do insurance companies decide what to cover?
- How can we make a difference?
- When will we be able to stop writing LMNs?
Presenters

- Robert Nussbaum, MD
  CMO, Invitae, Professor UCSF

- Katherine Spoonamore, MS, LCGC
  Genetic Counselor, IU School of Medicine
  Krannert Institute of Cardiology

- Brent J. O’Connell, MD, MHSA
  President and CMO of Christopher Place
  Health Care Solutions

- Amber Trivedi, MS, CGC
  Senior Vice President Informed DNA
  Provider and Client Services
Insurance Coverage: The GC Experience in a Cardiovascular Genetics Clinic

NSGC 34th Annual Education Conference
24 October 2015

Katie Spoonamore, MS, CGC, LGC
Indiana University School of Medicine
Disclosures

Clinical genetic counselor in academic setting, research funding

Invitae 2015 Cardio Advisory Board
Genetic Testing in a Cardiology Clinic

SETTING THE STAGE
Cardiovascular Genetics Clinic

THE GENETIC TESTING

- Always been panel-based
- TATs historically very long
  - Results not available to impact immediate patient care
  - Often focused on making surveillance plans for family members
- Historically not covered by insurance, cost prohibitive for patients

THE DISEASES

- Usually AD, less commonly AR, X-linked, mitochondrial
- Variable expressivity, age-of-onset
- Reduced penetrance
- Early diagnosis improves outcomes
COMMON INDICATIONS

CARDIOMYOPATHY

- Can impact decisions about devices, transplant
- 3% of HCM patients with GT found to have undetected syndromic disease\(^1\)
- Significantly different differentials in pediatric vs. adult populations

ARRHYTHMIA

- Can impact decisions about devices, transplant
- Can target high risk scenarios, medications to avoid

AORTOPATHY

- Can impact imaging strategy for surveillance
- Can impact surgical decision making
Guidelines for Genetic Testing in Cardiology

GUIDANCE FOR PRACTICING CLINICIANS
Who should have cardiac genetic testing?

AT-RISK RELATIVES

For relatives: Class I (is recommended)

Familial testing following the identification of a pathogenic mutation:
- LQTS
- CPVT
- Brugada
- Progressive CCD
- SQTS
- HCM
- ARVC
- DCM
- LVNC
- RCM
- SUD/SIDS

HRS/EHRA Expert Consensus Statement on the State of Genetic Testing for the Channelopathies and Cardiomyopathies

This document was developed as a partnership between the Heart Rhythm Society (HRS) and the European Heart Rhythm Association (EHRA)

Michael J. Ackerman, MD, PhD¹, Silvia G. Priori, MD, PhD², Stephan Willems, MD, PhD³, Charles Berul, MD, FHRSc, CCDS⁴, Ramon Brugada, MD, PhD⁵, Hugh Calkins, MD, FHRS, CCDS⁶, A. John Camm, MD, FHRS⁷, Patrick T. Ellinor, MD, PhD⁸, Michael Gollob, MD⁹, Robert Hamilton, MD, CCDS¹⁰, Ray E. Hershberger, MD¹¹, Daniel P. Judge, MD¹²,¹³, Hervé Le Marc, MD¹³, William J. McKenna, MD¹⁴, Eric Schulze-Bahr, MD, PhD¹⁵, Chris Semsarian, MBBS, PhD¹⁶, Jeffrey A. Towbin, MD¹⁷, Hugh Watkins, MD, PhD¹⁸, Arthur Wilde, MD, PhD¹⁹, Christian Wolpert, MD²⁰, and Douglas P. Zipes, MD, FHRS²¹

¹From Mayo Clinic, Rochester, Minnesota. ²Fontanese Salerno, Magenta University of Pavia, Pavia, Italy and New York University, New York, New York. ³University Hospital Hamburg-Eppendorf, Hamburg, Germany. ⁴Children’s National Medical Center and George Washington School of Medicine, Washington, District of Columbia. ⁵Cirque Institute of Biomedical Research and University of Girona School of Medicine, Girona, Spain. ⁶Johns Hopkins University, Baltimore, Maryland. ⁷St. George’s University of London, London, United Kingdom. ⁸Massachusetts General Hospital, Cardio Arrhythmia Service, Boston, Massachusetts. ⁹University of Ottawa Heart Institute, Ottawa, Canada. ¹⁰Hospital for Sick Children, Toronto, Canada. ¹¹University of Miami Miller School of Medicine, Miami, Florida. ¹²Salernitana Piansianese Crucciani, Pisa, Italy. ¹³Hôpital Saint-Denis, Nancy, France. ¹⁴Institute of Cardiovascular Science, University College London, London, United Kingdom. ¹⁵University Hospital Münster, Münster, Germany. ¹⁶University of Sydney, Sydney, Australia. ¹⁷Cincinnati Children’s Hospital, Cincinnati, Ohio. ¹⁸University of Oxford, John Radcliffe Hospital, Oxford, United Kingdom. ¹⁹University of Amsterdam Academisch Medisch Center, Amsterdam, The Netherlands. ²⁰Ludwigshafen Clinic, Ludwigshafen, Germany; and ²¹Kerckhoff Institute of Cardiology, Indiana University School of Medicine, Indianapolis, Indiana.
Who should have cardiac genetic testing?

### PROBAND/INDEX CASE

For a proband/index case: Class I (is recommended)

<table>
<thead>
<tr>
<th>Disease</th>
<th>Detection Rate</th>
</tr>
</thead>
<tbody>
<tr>
<td>LQTS</td>
<td>75-80%</td>
</tr>
<tr>
<td>(strong clinical index of suspicion established, or asymptomatic pt. w/ idiopathic QT prolongation of QTc&gt;480 ms prepuberty or QTc&gt;500 ms adults)</td>
<td></td>
</tr>
<tr>
<td>CPVT</td>
<td>70%</td>
</tr>
<tr>
<td>HCM</td>
<td>50-60%</td>
</tr>
<tr>
<td>DCM</td>
<td>&gt;20%</td>
</tr>
<tr>
<td>(w/ significant CCD and/or FHx of SUD)</td>
<td></td>
</tr>
<tr>
<td>Sudden Unexpected Death Cases</td>
<td>15% for SIDs (&lt;1 y.o.)</td>
</tr>
<tr>
<td>(with suspicion of cardiomyopathy or arrhythmia)*</td>
<td>25-35% for 1-35 y.o.</td>
</tr>
</tbody>
</table>

*Requires communication with medical examiners office to acquire appropriate specimens for testing
Who should have cardiac genetic testing?

PROBAND/INDEX CASE

HRS/EHRA Expert Consensus Statement on the State of Genetic Testing for the Channelopathies and Cardiomyopathies

This document was developed as a partnership between the Heart Rhythm Society (HRS) and the European Association (EHRA)

For a proband/index case: Class IIa (can be useful)

<table>
<thead>
<tr>
<th>Disease</th>
<th>Detection Rate</th>
</tr>
</thead>
<tbody>
<tr>
<td>Brugada syndrome type 1</td>
<td>25-35%</td>
</tr>
<tr>
<td>ARVC (when task force diagnostic criteria are met)</td>
<td>30-70%</td>
</tr>
<tr>
<td>DCM (familial cases)</td>
<td>&gt;20%</td>
</tr>
<tr>
<td>LVNC</td>
<td>15-20%</td>
</tr>
</tbody>
</table>
Who should know about changes in cardiovascular disease genes?

EVERYONE UNDERGOING CLINICAL WES, WGS

ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing

Robert C. Green, MD, MPH,1,2, Jonathan S. Berg, MD, PhD,1 Wayne W. Grody, MD, PhD,1,6, Sarah S. Kalia, ScM, CGC,1 Bruce R. Korf, MD, PhD,2, Christa L. Martin, PhD, FACMG,3, Amy L. McGuire, JD, PhD, Robert L. Nussbaum, MD,10, Janianne M. O'Daniel, MS, CGC,1 Kelly E. Ormond, MS, CGC,11, Heidi L. Rehm, PhD, FACMG,12, Michael S. Watson, PhD, FACMG,13, Marc S. Williams, MD, FACMG,14 and Leslie G. Biesecker, MD15

Disclaimer: These recommendations are designed primarily as an educational resource for medical geneticists and other health-care providers to help them provide quality medical genetic services. Adherence to these recommendations does not necessarily ensure a successful medical outcome. These recommendations should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, geneticists and other clinicians should apply their own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these recommendations.
Who should know about changes in cardiovascular disease genes?

<table>
<thead>
<tr>
<th>Phenotype</th>
<th>MIM-disorder</th>
<th>PMID-Gene Reviews entry</th>
<th>Typical age of onset</th>
<th>Gene</th>
<th>MIM-gene</th>
<th>Inheritance</th>
<th>Variants to report</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypertrophic cardiomyopathy, dilated cardiomyopathy</td>
<td>115197, 192600, 601494, 613690, 115196, 608751, 612098, 600858, 301500, 608758, 115200</td>
<td>20301725</td>
<td>Child/adult</td>
<td>MYBPC3</td>
<td>600958</td>
<td>AD</td>
<td>KP and EP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>MYH7</td>
<td>160760</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>TNNT2</td>
<td>191045</td>
<td></td>
<td>KP and EP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>TNNT3</td>
<td>191044</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>TPM1</td>
<td>191010</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>MYL3</td>
<td>160790</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>ACTC1</td>
<td>102540</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>PRKAG2</td>
<td>602743</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>GLA</td>
<td>300644</td>
<td>XL</td>
<td>KP and EP (hemi, het, hom)</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>MYL2</td>
<td>160781</td>
<td>AD</td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>LMNA</td>
<td>150330</td>
<td>AD</td>
<td>KP</td>
</tr>
<tr>
<td>Catecholaminergic polymorphic ventricular tachycardia</td>
<td>604772</td>
<td></td>
<td></td>
<td>RYR2</td>
<td>180902</td>
<td>AD</td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>PKP2</td>
<td>602861</td>
<td>AD</td>
<td>KP and EP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>DSP</td>
<td>125647</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>DSC2</td>
<td>125645</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>TMEM43</td>
<td>612048</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>DSG2</td>
<td>125671</td>
<td></td>
<td>KP and EP</td>
</tr>
<tr>
<td>Arrhythmogenic right-ventricular cardiomyopathy</td>
<td>609040, 604400, 610476, 607450, 610193</td>
<td>20301310</td>
<td>Child/adult</td>
<td>PKP2</td>
<td>602861</td>
<td>AD</td>
<td>KP and EP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>MKK3</td>
<td>125671</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>TMEM43</td>
<td>612048</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>DSG2</td>
<td>125671</td>
<td></td>
<td>KP and EP</td>
</tr>
<tr>
<td>Romano–Ward long QT syndrome types 1, 2, and 3, Brugada syndrome</td>
<td>192500, 613688, 603830, 601144</td>
<td>20301308</td>
<td>Child/adult</td>
<td>KCNQ1</td>
<td>607542</td>
<td>AD</td>
<td>KP and EP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>KCNH2</td>
<td>152427</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>SCN5A</td>
<td>600163</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td>Familial hypercholesterolemia</td>
<td>143890, 603776</td>
<td>No GeneReviews entry</td>
<td>Child/adult</td>
<td>LDLR</td>
<td>606945</td>
<td>SD</td>
<td>KP and EP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>APOB</td>
<td>107730</td>
<td>SD</td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>PCSK9</td>
<td>607786</td>
<td>AD</td>
<td>KP</td>
</tr>
<tr>
<td>Ehlers–Danlos syndrome, vascular type</td>
<td>130050</td>
<td>20301667</td>
<td>Child/adult</td>
<td>COL3A1</td>
<td>120180</td>
<td>AD</td>
<td>KP and EP</td>
</tr>
<tr>
<td>Marfan syndrome, Loeys–Dietz syndromes, and familial thoracic aortic aneurysms and dissections</td>
<td>154700, 609192, 608967, 610168, 610380, 613795, 611788</td>
<td>20301510, 20301312, 20301299</td>
<td>Child/adult</td>
<td>FBN1</td>
<td>134797</td>
<td>AD</td>
<td>KP and EP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>TGFBR1</td>
<td>190181</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>TGFBR2</td>
<td>190182</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>SMAD3</td>
<td>603109</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>ACTA2</td>
<td>102620</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>MYLK</td>
<td>600922</td>
<td></td>
<td>KP</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>MYH11</td>
<td>160745</td>
<td></td>
<td>KP</td>
</tr>
</tbody>
</table>
Challenges with Guidelines:

- Don’t reflect actual use of genetic testing in clinical practice, nonspecific
- Not regularly updated, quickly outdated
- Rarely consistent with insurance policies regarding coverage
Insurance Policies

GUIDANCE FOR PAYERS, BY PAYERS
Why these genes?

No access to bigger panels?
And/or no reimbursement for bigger panels?

No coverage for those w/ clear diagnosis, to determine subtype and management implications?

LQTS: Medically necessary in patients who DO NOT meet diagnostic criteria for LQTS (Schwartz <4), or in at-risk family members with a known mutation in the family

LQTS: Only covered in at-risk family members when there is a known LQT mutation and proband has had SCD, syncope or VF.

Why these genes?

No access to bigger panels?
And/or no reimbursement for bigger panels?

No coverage for those w/ clear diagnosis, to determine subtype and management implications?

LQTS: Medically necessary in patients who DO NOT meet diagnostic criteria for LQTS (Schwartz <4), or in at-risk family members with a known mutation in the family

LQTS: Only covered in at-risk family members when there is a known LQT mutation and proband has had SCD, syncope or VF.

No coverage if proband’s presentation wasn’t severe enough?
Lab Billing Policies
Lab Billing Policies

- Greatly increased access to testing in cardiology
- Decreased real, perceived barriers for patients, cardiologists
- Distances GC from billing process (for better, for worse)
Challenges to Caring For Patients in Clinic

NAVIGATING THE ROAD TO GENETIC TESTING
Challenges in Clinic

- Disconnect between practice guidelines, coverage policies

- Can a patient get testing? Today?
  - Insurance? (commercial, Medicare, Medicaid, none?)
  - Billing capabilities? (institutional?)

- Cost? GC job to determine, communicate?

- Adjust testing strategy?

- How do we talk about this with patients in session?
Clinical Genetic Counselor Experience

Blissfully Unaware of Billing Details

- Only hear about specific situations when it’s a problem
- Not truly sure what’s covered or not
- Rarely have problems with patients getting needed genetic testing

Hyperaware of Ever-Changing Billing Details

- Regularly perform pre-auths, write LMNs, do peer-to-peers
- Up-to-date with specific, changing policies
- Often delay patient’s genetic testing to complete insurance processes
- Sometimes have patients who are denied, cannot get testing
“Genesurance Counseling”

**Verb** \([\text{JEEN-S'HOO R-UH NSS}] \ [\text{KOUN-SUH-LING}]\) : That portion of a genetic counseling session, whether intentional or non-intentional, that is devoted to the topic of costs and insurance/3rd party coverage (particularly for genetic testing).
STUDENT RESEARCH SURVEY: GENESURANCE COUNSELING

GENESURANCE COUNSELING

VERB [JEEN-SHOO R-UH NSS] [KOUN-SUH-LING]

: THAT PORTION OF A GENETIC COUNSELING SESSION, WHETHER INTENTIONAL OR NON-INTENTIONAL, THAT IS DEVOTED TO THE TOPIC OF COSTS AND INSURANCE/3RD PARTY COVERAGE (PARTICULARLY FOR GENETIC TESTING).

ARE YOU A GENETIC COUNSELOR WHO COUNSELS PATIENTS ON A REGULAR BASIS? WE WANT TO HEAR FROM YOU ABOUT YOUR WORKDAY, SPECIFICALLY ABOUT YOUR EXPERIENCE WITH FINANCIAL/INSURANCE RELATED TOPICS AND HOW THAT MAY IMPACT THE WAY YOU COUNSEL PATIENTS. THIS SURVEY AIMS TO DETERMINE HOW MUCH TIME GENETIC COUNSELORS ARE SPENDING DISCUSSING AND RESEARCHING INSURANCE RELATED CONCEPTS, AND IF THIS RESPONSIBILITY HAS INCREASED OVER TIME. THIS SURVEY IS COMPOSED OF 20 QUESTIONS AND WILL TAKE ROUGHLY 10-15 MINUTES TO COMPLETE. IF YOU CONSENT TO PARTICIPATE IN THIS SURVEY YOU WILL HAVE THE OPTION OF BEING ENTERED INTO A RANDOM DRAWING FOR 1 OF 5 $20.00 AMAZON GIFT CARDS. IF YOU WISH TO COMPLETE THIS SURVEY PLEASE CLICK THE LINK BELOW.

SURVEYMONKEY

THIS STUDY HAS BEEN APPROVED BY THE SANFORD HEALTH IRB (ID STUDY00000423 )

SHELBY BROWN
GENETIC COUNSELING STUDENT
UNIVERSITY OF ARKANSAS FOR MEDICAL SCIENCES
SBROWN4@UAMS.EDU

QUINN STEIN, MS, CGC
LICENSED GENETIC COUNSELOR
SANFORD HEALTH
QUINN.STEIN@SANFORDHEALTH.ORG
STUDENT RESEARCH SURVEY: GENESURANCE COUNSELING

GENESURANCE COUNSELING

*Verb [JEEN-SHOO R-UH NSS] [KOUN-SUH-LING]*

That portion of a genetic counseling session, whether intentional or non-intentional, that is devoted to the topic of costs and insurance/third party coverage (particularly for genetic testing).

Are you a genetic counselor who counsels patients on a regular basis? We want to hear from you about your workday, specifically about your experience with financial/insurance related topics and how that may impact the way you counsel patients. This survey aims to determine how much time genetic counselors are spending on insurance related concepts, and if this responsibility has increased over time.

This survey is composed of 20 questions and will take roughly 10-15 minutes to complete. If you consent to participate in this survey you will have the option of being entered into a random drawing for 1 of 5 $20.00 Amazon gift cards. If you wish to complete this survey please click the link below.

SurveyMonkey

This study has been approved by the Sanford Health IRB (ID STUDY00000423)

Shelby Brown
Genetic Counseling Student
University of Arkansas for Medical Sciences
SBROWN4@UAMS.EDU

Quinn Stein, MS, CGC
Licensed Genetic Counselor
Sanford Health
QUINN.STEIN@SANFORDHEALTH.ORG

Nearly 400 people have completed the survey already

99.21% say they do genesurance counseling as part of their GC session

Currently spending an average of 7-8 minutes discussing genesurance counseling
Thoughts for the future
Ways to spend our time and energy:

- Should a family member’s policy pay for the testing in some scenarios?

- How can we engage with the process to continue increasing ease for patients and providers, while also helping to create sustainable change?

- From a health economics standpoint, genetic testing can provide cost savings.\(^1,2,3\) We need more data!

- We need consistently updated, specific guidelines backed by appropriate cardiology and genetics societies (HRS, HFSA, ACC, NSGC, ACMG)

- New clinical relationships to increase multidisciplinary care to encourage appropriate use of guidelines, reduce inappropriate testing, etc.
Appropriate Coverage of Genetic Testing

- Low-Cost for Patients
- Ease of Use for Clinicians
- Sustainable Access
Thank You

KATIE SPOONAMORE, MS, CGC, LGC
CERTIFIED, LICENSED GENETIC COUNSELOR
KRANNERT INSTITUTE OF CARDIOLOGY
INDIANA UNIVERSITY SCHOOL OF MEDICINE
IU HEALTH PHYSICIANS CARDIOLOGY
KSPOONAM@IU.EDU

The work described was supported by the Indiana University Health-Indiana University School of Medicine Strategic Research Initiative and the Indiana Genomics Initiative.
Ever feel like you are alone?
Understanding Insurance

SUGGESTIONS ON APPROACHING PAYERS
Insurance coverage

- Multiple players
  - Medicare
  - Blues
  - Commercials
  - Medicaid
  - Self Insured

1. Different Rules for each
2. Different legislation governs each type
Issues for Genetic Counselors

SEVERAL ISSUES
Issues for genetic counselors

1. Does the health plan recognize a genetic counselor

2. DO THEY CREDENTIAL GENETIC COUNSELORS
   1. MS or doctoral degree
   2. AMGC or ABMG accredited
   3. Passed the exams
   4. The process can take months to complete
   5. In network or not? Big difference
What do they allow genetic counselors to do?

1. Office visits/consultations
2. Ordering testing
3. Order treatment

How many does a plan need?
Investigational services

vs

Not medically necessary

MAPLE SYRUP URINE DISEASE
TRISOMY 21
CYSTIC FIBROSIS
How to change what you see as injustice
Have you ever visited a medical director?  
Do you know who he is?  
Ever mailed a medical director?  
Ever asked to be a consultant?
Talk to the correct person

- Chief Medical Officer
- Medical Policy physicians
- Utilization management physicians
- Quality physicians
- Pre-authorization physicians
- Provider relations physicians
- Consultants
Parting comments

1. Medical directors may disagree with you. Find out why!
2. Do not be abusive! Makes things worse.
3. Offer your services for education
4. Better to meet the medical director when you don’t have an issue
There is light at the end

THANK YOU
Amber Trivedi MS, CGC
Senior VP, Provider & Client Services
InformedDNA

DISCLOSURE
Genetic Benefit Management: Following the xBM model

1980: Pharmacy Benefit Management (PBM)
1990: Radiology Benefit Management (RBM)
2000: Genetic Benefit Management (GBM)
2010:
2020:

Right Patient
Right Test
Right Lab
Right Decisions
Genetic Benefit Management Services

- Policy development
- Review prior auth requests and claims for medical necessity
- Outreach to ordering providers
  - Obtain additional clinical information
  - Discuss rationale and impact on medical management
  - Explore alternate testing options

Comprehensive Genetic Benefits Management

- Reporting & Measurement
- Policies & Guidelines
- Data-Driven UM Program
- Genetic Clinical Expertise
Different perspectives, but a common goal

**GENETIC COUNSELORS**

- Focus on providing the most comprehensive care for the individual
- How can I be an advocate for my patient and get him/her the tests s/he desires?
- Will the test results yield new information for my patient?

**PAYERS**

- Focus on providing equitable and cost-effective care across an entire population
- Should this test be ordered for every patient with the same clinical scenario?
- Will the test results lead to a change in management?

*Both want to provide excellent healthcare services with the goal of improved patient outcomes*
Medically Necessary: What does that really mean?

**Medical necessity (aka clinical utility): evidence shows that test is likely to improve patient outcomes**

**Factors payers often DO consider medically necessary**
- Results are likely to lead to a change in medical management
- Results are likely to lead to increased surveillance
- Results are likely to impact reproductive decision-making
- Results are likely to identify agents or circumstances to avoid

**Factors payers often DO NOT consider medically necessary**
- Results yield info for which there are no established management guidelines
- Screening tests with low positive predictive value
- Results will inform a research question or academic curiosity
- Results may have some psychological impact but no impact on medical management
Experimental, investigational, or unproven: test does not have evidence-based medical necessity

**Experimental DOES mean**
- Test doesn’t meet the definition of medically necessary in any clinical scenario
- Insufficient evidence in the literature that test results will improve patient outcomes for the specific indication/presenting features

**Experimental DOES NOT mean**
- It’s a bad test
- It’s a test that’s still undergoing development/research
- It’s a test with no data behind it
Cost-Effective: What does that really mean?

**Cost-effectiveness analysis:**

**(costs with testing – costs without testing)**

**(health benefit with testing – health benefit without testing)**

Cost-effectiveness **DOES** mean:

- Using a specific calculation to compare one intervention to another
- Using an accepted scientific measurement, e.g. quality-adjusted life year (QALY)

Cost-effective **DOES NOT** mean:

- A cheaper test
- A more efficient test
- Looking at test cost without factoring in outcomes
Changing landscape of evidence review

- CDC ACCE framework is gold standard for evaluating genomic tests
- Challenges to this framework for NGS
  - Can assess ACCE for individual genes, but difficult to assess a panel where genes have varying levels of evidence
  - Standard has been to call whole panel investigational unless every gene had robust evidence
- Payers starting to recognize these challenges and are seeking ways to reconcile
Changing landscape of evidence review

_How do you systematically factor in the “E” in ACCE?_

- How does cost effectiveness fit into the equation?
- What about risk of harm?
- Will result lead to procedures that pose risk to the patient yet are not evidence-based for that indication?
- Will result lead to use of treatment that is not proven for that indication?
Example: whole exome sequencing

**Most payers still consider WES as investigational, but some have recognized clinical utility for certain scenarios**

- Tests without evidence describing the results expected for a specific indication are perceived as “a fishing expedition” or “throwing spaghetti at the wall to see what sticks”
  - Payers view this as research testing
  - Evidence should be specific about outcomes for a given indication, not vague or broad

- ACMG defines practical use of WES, but referencing guidelines is too broad to address clinical utility for a specific case

- Clinicians can clarify that WES is practical by:
  - Providing a detailed differential diagnosis list
  - Specifying potential management changes yielded by WES
  - Describing tests/procedures that would be ordered in the absence of WES
Example: whole exome sequencing

- 5yo female with developmental delays and slow growth. Brain MRI with demyelination and Dandy-Walker variant with a degree of bilateral inferior cerebellar hypoplasia. Patient is microcephalic but otherwise non-dysmorphic. Previous testing included metabolic work-up with primary finding of reduced arylsulfatase activity which is indicative of either carrier status for a mutation in the arylsulfatase gene or pseudoaryl sulfatase A deficiency.

- Large lysosomal storage panel identified 4 VUSs. Microarray was normal.

- No additional conditions on the differential that had not already been tested for

- WES considered investigational for this clinical scenario
Example: whole exome sequencing

- 5yo male with hypotonia, cardiomyopathy, intellectual disability and autism.
- Normal chromosomes, CMA, fragile X, metabolic studies, and Prader-Willi/Angelman testing.
- Ordering provider documented:
  - A specific list of 9 conditions remaining on the differential. Described homogeneous presentation of those conditions and inability to perform sequential testing based on likelihood of disorder. Described how WES was more cost effective than testing for the 9 conditions individually.
  - Results were expected to direct cardiology management and consideration of heart transplant, and reproductive decision-making for the family.
- WES considered medically necessary for this clinical scenario
Communicating with payers

*When writing LMNs or communicating via phone…*

<table>
<thead>
<tr>
<th><strong>DO</strong></th>
<th><strong>DON’T</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>• Provide concrete examples of how test will change management for the specific patient</td>
<td>• Make vague or broad statements about medical necessity of test in general without providing case-specific details</td>
</tr>
<tr>
<td>• Include case-specific details and rationale for testing</td>
<td>• Reference cost-effectiveness without providing details (e.g. large number of genes on differential diagnosis list)</td>
</tr>
<tr>
<td>• Cite evidence in the literature to support testing or medical management changes</td>
<td></td>
</tr>
<tr>
<td>• Reference national and professional guidelines</td>
<td></td>
</tr>
</tbody>
</table>
Panel Q&A and Discussion