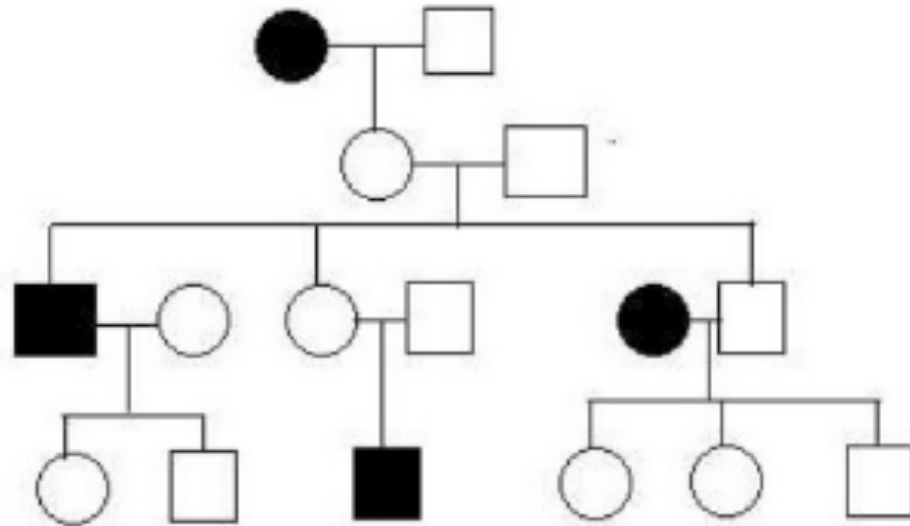


Challenges Related to Family Involvement in Clinical Whole-Genome Sequencing: Views of Non-Genetics Providers

Leila Jamal, ScM, PhD, CGC
For The MedSeq Project



Family Communication Matters

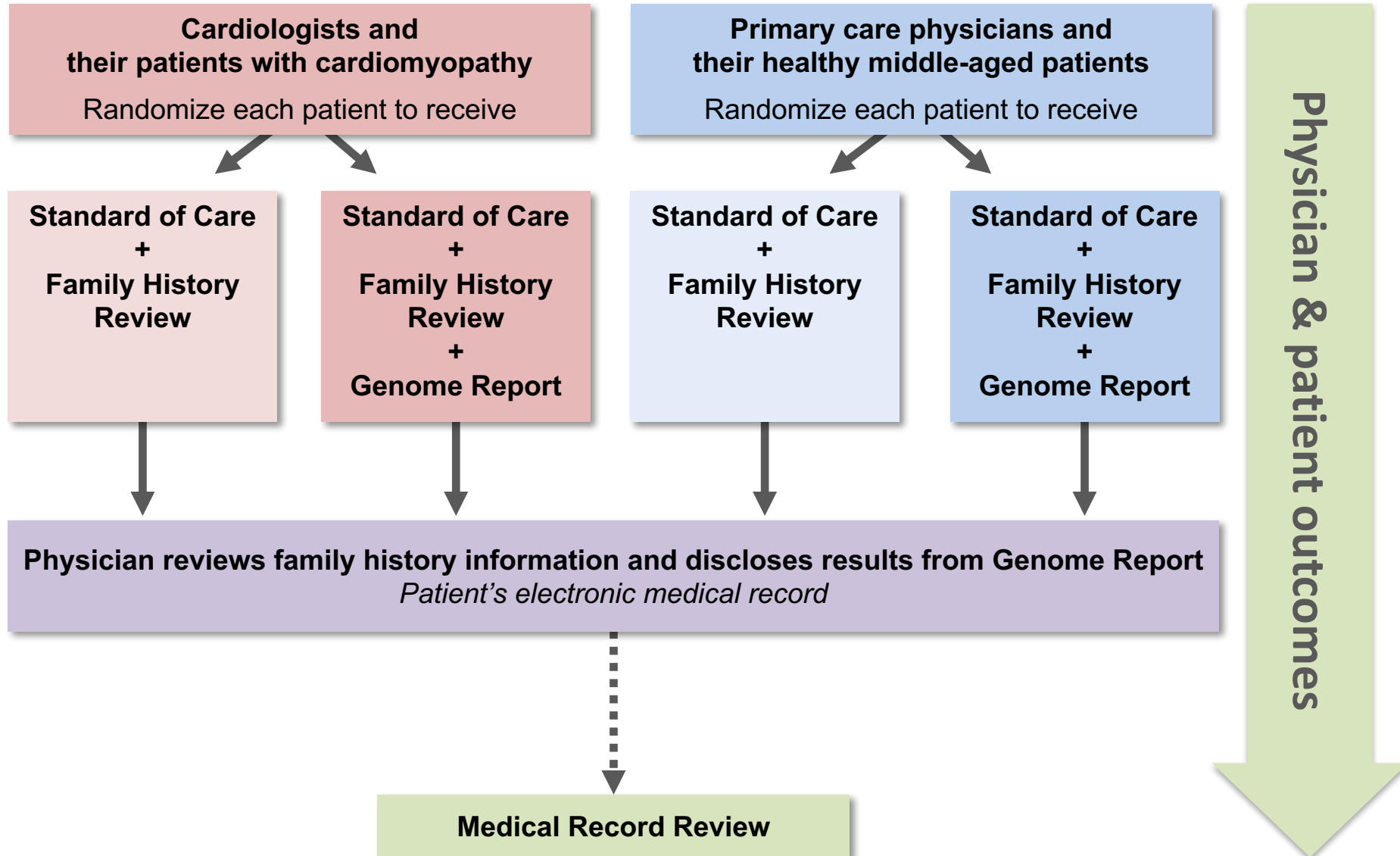


Communicating Genetic Information to Patients' Families

- New issue for non-genetics providers
- Guidelines
 - Discourage providers from contacting relatives directly
 - Encourage providers to help patients to transmit risk information to relatives
 - Little clarity about how
 - Differ for research and clinical spheres

The MedSeq Project

U01 HG006500 (2012-2016)



Patient and Physician Demographics

Characteristic (%) unless noted	Patients (n = 205)	Physicians (n = 20)
Mean age (sd)	55.2 (11.4)	51.7 (10.0)
Age range	19 – 85	32 – 65
Gender		
Female	104 (51%)	8 (40%)
Male	101 (49%)	12 (60%)
Race		
Non-Hispanic white	177 (86%)	15 (75%)
Other/No response	28 (14%)	5 (25%)
Annual Household Income		
<\$100,000	71 (35%)	-
≥\$100,000	125 (61%)	-
Education		
Did not graduate from college	38 (19%)	0 (0%)
College graduate or higher	166 (81%)	100 (100%)

Patients' Attitudes Re: Family Involvement

“The most important thing about this was to prevent something, my children’s future... that was what I was concerned about more, that this study might prevent something from happening to them.” (186-P05)

Information Sharing		PD surveys		6 month	
		Control (n=87)	WGS (n = 97)	Control (n = 75)	WGS (n = 91)
My spouse or partner	Yes	75%	78%	72%	78%
	No	11%	9%	12%	9%
If yes, what type of information?					
General information and/or feelings about my info		94%	97%	96%	96%
My risk and/or my relative's risk of having a condition		40%	63%	28%	46%
Possibility of employment or insurance discrimination		2%	5%	6%	6%
Preventative surgery and/or screening recommendations		18%	36%	20%	20%
My child(ren)	Yes	52%	63%	39%	53%
	No	25%	14%	36%	29%
If yes, what type of information?					
General information and/or feelings about my info		87%	87%	90%	79%
My risk and/or my relative's risk of having a condition		40%	52%	48%	56%
Possibility of employment or insurance discrimination		2%	10%	0%	2%
Preventative surgery and/or screening recommendation		31%	36%	31%	17%
Siblings	Yes	54%	72%	41%	58%
	No	28%	21%	39%	30%
If yes, what type of information?					
General information and/or feelings about my info		91%	93%	90%	91%
My risk and/or my relative's risk of having a condition		51%	66%	61%	60%
Possibility of employment or insurance discrimination		0%	6%	3%	2%
Preventative surgery and/or screening recommendation		36%	37%	42%	32%

Bold indicates majority response. **Green** indicates notable increase in WGS compared to Control.

Familial risk assessment was an unexpected issue

“I hadn’t been thinking about when I need to worry about somebody having an autosomal recessive trait. What about the rest of their family? I mean, it wasn’t even on my radar.” -P01

MDs viewed family communication as patient responsibility

“It’s the equivalent of knowing your patient is using drugs, and you’d like to tell their spouse. You can’t, unless they’re going to hurt somebody. There’s going to be very strict guidelines for how this information can be released to relatives...we just don’t do things that way. It would be nice if we could, but we can’t.” –P10

MDs viewed family communication as patient responsibility

“It's hard to know where that line is...if it's some life-threatening situation, or a condition where early intervention makes a difference, it's hard for the physician not to be able to directly approach family members. But right now, there's no avenue for that.” -C02

Special circumstances might make it ok to contact family members

“In the event of death where there was no prior knowledge of the person's wishes, I think that would be a very different scenario. For something that had ramifications that we could be certain had a high probability of happening, I would reach out first to the person's spouse...” –C06

MDs expressed a need for tools

“I think it would be helpful to have something in print for giving to family members. It's a very weird situation because you are kind of getting involved in the care of people you have never laid eyes on, and never will.” – P14

“I think there must be ways you can share risk without necessarily sharing specific genetic information about an individual” –C08

Approaches to sharing genetic info with relatives

- Group information sessions with voluntary patient follow up
- Telephone counseling/Telemedicine
- Prospective consent to contact relatives obtained from index patient

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Summary

- Family involvement in WGS may be a novel challenge for non-genetics providers
- Important throughout the process, but our MDs focused most on sharing results
- Different approaches for different result types?

The MedSeq Project Collaborators

Project Leadership

Robert Green, MD, MPH
Zak Kohane, MD, PhD
Calum MacRae, MD, PhD
Amy McGuire, JD, PhD
Michael Murray, MD
Heidi Rehm, PhD
Christine Seidman, MD
Jason Vassy, MD, MPH, SM

Project Manager

Carrie Blout, MS

Project Personnel

Sandy Aronson, ALM, MA
Danielle Azzariti, MS
David Bates, MD
Jennifer Blumenthal-Barby, PhD
Ozge Ceyhan-Birsoy, PhD
Alexis Carere, MA, MS, PhD
Kurt Christensen, MPH, PhD
Allison Cirino, MS
Lauren Conner
Kelly Davis, PhD

Project Personnel (Cont.)

Dmitry Dukhovny, MD, MPH
Lindsay Feuerman
Margaret Helm, MS
Carolyn Ho, MD
Lily Hoffman-Andrews
Peter Kraft, PhD
Joel Krier, MD
Sek Won Kong, MD
William Lane, MD, PhD
Matt Lebo, PhD
Lisa Lehmann, MD, PhD, MSc
In-Hee Lee, PhD
Kaitlyn Lee
Kalotina Machini, PhD, MS
David Margulies, MD
Heather McLaughlin, PhD
Jill Robinson, MA
Melody Slashinski, MPH, PhD
Shamil Sunyaev, PhD
Ellen Tsai, PhD
Peter Ubel, MD
Rebecca Walsh
Scott Weiss, MD

External Advisory Board

Katrina Armstrong, MD
David Bentley, DPhil
Robert Cook-Deegan, MD
Muin Khoury, MD, PhD
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Jim Lupski, MD, PhD
Kathryn Phillips, PhD
Lisa Salberg
Maren Scheuner, MD, MPH
Sue Siegel, MS
Sharon Terry, MA

Consultants

Les Biesecker, MD
George Church, PhD
Geoffrey Ginsburg, MD, PhD
Tina Hambuch, PhD
David Miller, MD, PhD
J. Scott Roberts, PhD
David Veenstra, PharmD, PhD

Protocol Monitoring Committee

Judy Garber, MD, MPH
Cynthia Morton, PhD