

The Role and Impact of Personal Faith and Religion in the Lives of Genetic Service Providers

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**Genetic Counselor
Clinical Professor of Medicine**

Today's talk

- Genetic counseling
- Genetic testing, including new technologies
- Findings from our “Distress and Suffering of Genetic Services Providers” project
- Role and impact of personal faith and religion in the lives of genetic counselors

What is genetic counseling?

“Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease”



- ❑ Master's trained professionals
- ❑ Generally women
- ❑ ~2500 nationally
- ❑ Licensed in ~18 states

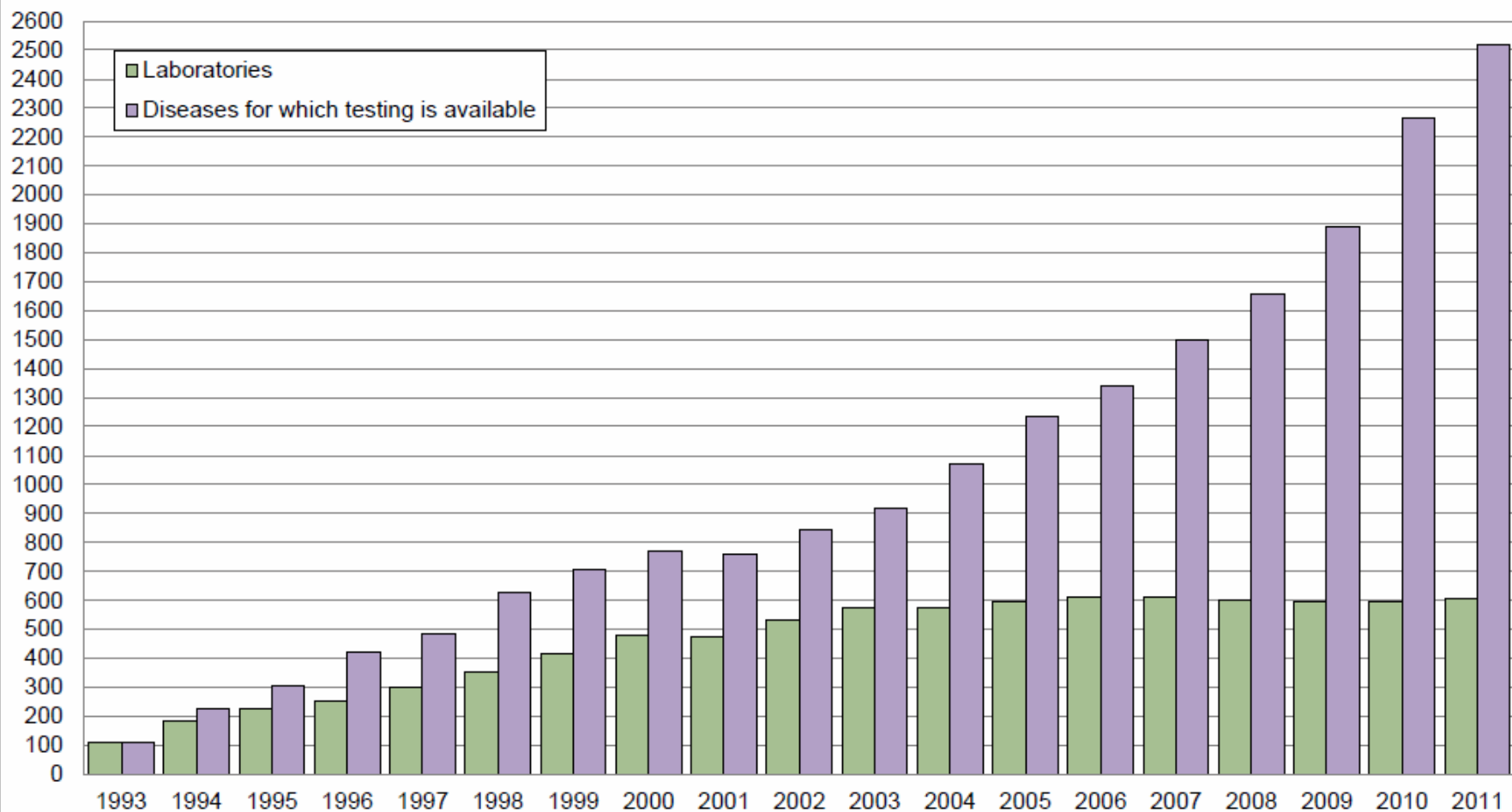
Genetic counselor practice settings

- Prenatal
 - Couples at increased risk for genetic disorders/birth defects due to abnormal maternal screening tests, family history, maternal age, etc.
- Pediatric
 - Children with multiple birth defects or developmental delay
 - Children with known genetic conditions
- Adult
 - People with a personal or family history of genetic disorders or adult-onset disorders (general genetics, oncology, neurology, cardiology, etc)
- Public health
- Industry
- Direct-to-consumer genetic testing

What does genetic counseling involve?

- Assessing client knowledge, values, experiences and expectations
- Interpretation of family and medical histories (including genetic test results) to assess the chance of disease occurrence or recurrence
- Education about inheritance, testing, management, prevention, resources and research
- Counseling (usually non-directive) to provide support, and promote informed choice and adaptation to the risk or condition
- Discussion about the results and implications of genetic testing

GENE Tests: Growth of Laboratory Directory



Data source: GeneTests database (2011)/ www.genetests.org

Uses of clinical genetic tests

- For diagnosis based on symptoms

Coming to a clinic near you...

Use of Whole-Genome Sequencing to Diagnose a Cryptic Fusion Oncogene

John S. Welch, MD, PhD

Peter Westervelt, MD, PhD

Li Ding, PhD

David E. Larson, PhD

Jeffery M. Klee, MD, PhD

Shashikant Kulkarni, PhD

John Wallis, PhD

Ken Chen, PhD

Context Whole-genome sequencing is becoming increasingly available for research purposes, but it has not yet been routinely used for clinical diagnosis.

Objective To determine whether whole-genome sequencing can identify cryptic, actionable mutations in a clinically relevant time frame.

Design, Setting, and Patient We were referred a difficult diagnostic case of acute promyelocytic leukemia with no pathogenic *X-RARA* fusion identified by routine metaphase cytogenetics or interphase fluorescence in situ hybridization (FISH). The case patient was enrolled in an institutional review board–approved protocol, with consent specifically tailored to the implications of whole-genome sequencing. The protocol uses a “movable firewall” that maintains patient anonymity within the entire research team

Clinical assessment incorporating a personal genome

Evan A Ashley, Atul J Butte, Matthew T Wheeler, Rong Chen, Teri E Klein, Frederick E Dewey, Joel T Dudley, Kelly E Ormond, Aleksandra Pavlovic, Alexander A Morgan, Dmitry Pushkarev, Norma F Neff, Louanne Hudgins, Li Gong, Laura M Hodges, Dorit S Berlin, Caroline F Thom, Katrin Sangkuhl, Joan M Hebert, Mark Woon, Hersh Sagreiya, Ryan Whaley, Joshua W Knowles, Michael F Chou, Joseph V Thakuria, Abraham M Rosenbaum, Alexander Wait Zaranek, George M Church, Henry T Greedy, Stephen R Quake, Russ B Altman

Making a definitive diagnosis: Successful clinical application of whole exome sequencing in a child with intractable inflammatory bowel disease

Elizabeth A. Worthey, PhD^{1,2}, Alan N. Mayer, MD, PhD^{2,3}, Grant D. Syverson, MD², Daniel Helbling, BSc⁴, Benedetta B. Bonacci, MSc², Brennan Decker, BSc⁴, Jaime M. Serpe, BSc², Trivikram Dasu, PhD², Michael R. Tschannen, BSc⁴, Regan L. Veith, MSc², Monica J. Basehore, PhD⁴, Ulrich Broeckel, MD, PhD^{1,2,5}, Aoy Tomita-Mitchell, PhD^{1,2,5}, Marjorie J. Arca, MD^{3,5}, James T. Casper, MD^{2,3}, David A. Margolis, MD^{2,3}, David P. Bick, MD^{1,2,3}, Martin J. Hessner, PhD^{1,2}, John M. Routes, MD^{2,3}, James W. Verbsky, MD, PhD^{2,3}, Howard J. Jacob, PhD^{1,2,3,6}, and David P. Dimmock, MD^{1,2,3}

Identification of a Novel *TP53* Cancer Susceptibility Mutation Through Whole-Genome Sequencing of a Patient With Therapy-Related AML

Daniel C. Link, MD

Context The identification of patients with inherited cancer suscep

Genetic diagnosis by whole exome capture and massively parallel DNA sequencing

Murim Choi^a, Ute I. Scholl^b, Weizhen Ji^b, Tiewen Liu^b, Irina R. Tikhonova^b, Paul Zumbo^b, Ahmet Nayir^c, Ayşin Bakkalıoğlu^d, Seza Özen^d, Sami Sanjad^e, Carol Nelson-Williams^b, Anita Farhi^b, Shrikant Mane^b, and Richard P. Lifton^{a,1}

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Whole-Genome Sequencing in a Patient with Charcot–Marie–Tooth Neuropathy

James R. Lupski, M.D., Ph.D., Jeffrey G. Reid, Ph.D., Claudia Gonzaga-Jauregui, B.S., David Rio Deiros, B.S., David C.Y. Chen, M.Sc., Lynne Nazareth, Ph.D., Matthew Bainbridge, M.Sc., Huyen Dinh, B.S., Chyn Jing, M.Sc., David A. Wheeler, Ph.D., Amy L. McGuire, J.D., Ph.D., Feng Zhang, Ph.D., Pawel Stankiewicz, M.D., Ph.D., John J. Halperin, M.D., Chengyong Yang, Ph.D., Curtis Gehman, Ph.D., Danwei Guo, M.Sc., Rola K. Irikat, B.S., Warren Tom, B.S., Nick J. Fantin, B.S., Donna M. Muzny, M.Sc., and Richard A. Gibbs, Ph.D.

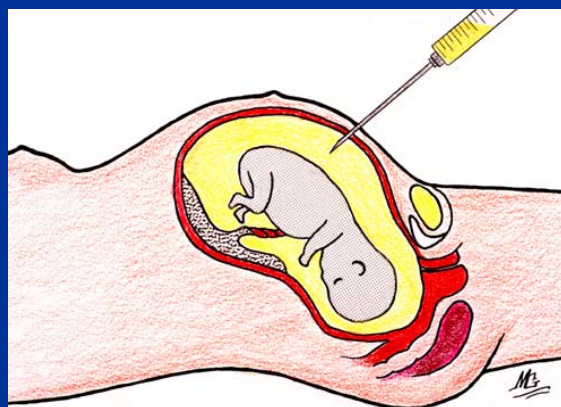
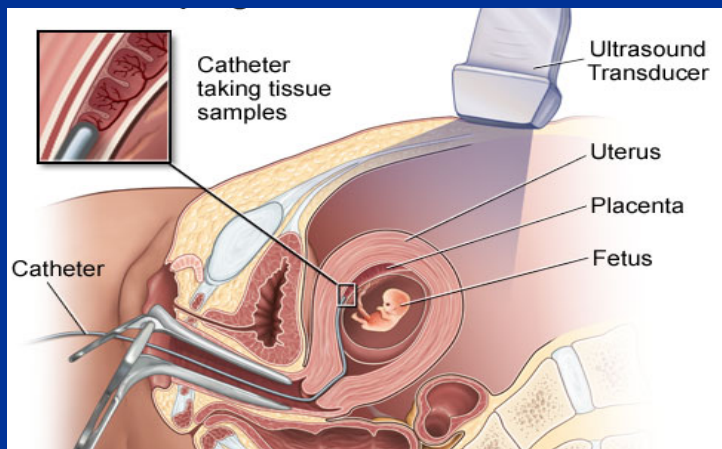
N Engl J Med 2010; 362:1181-1191 | April 1, 2010

Uses of clinical genetic tests

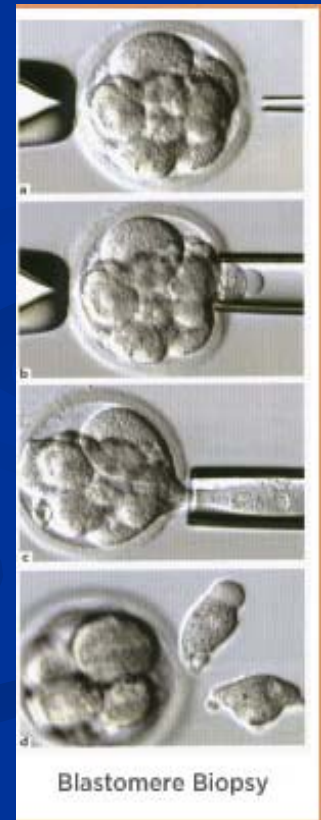
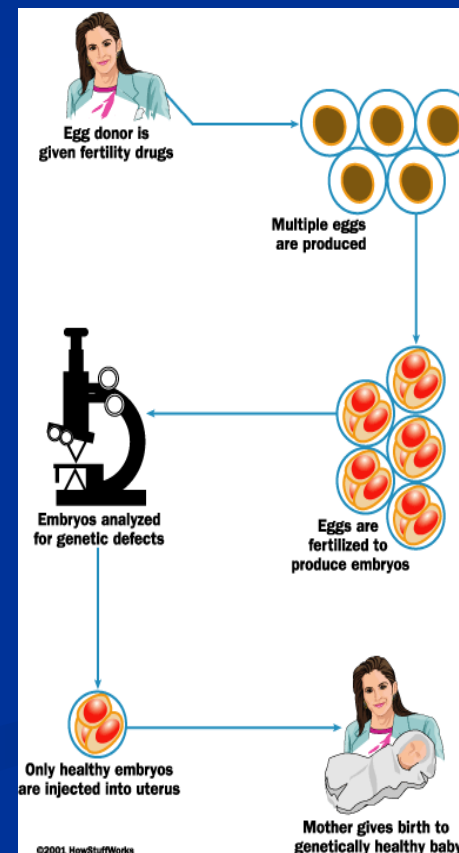
- For diagnosis based on symptoms
- Prenatal testing
- To predict risk
 - Presymptomatic testing (including newborn screening)
 - Prenatal screening
 - Carrier screening
 - Testing for susceptibility to common complex diseases
- To guide treatment
 - Somatic tumor testing
 - Pharmacogenetic testing
- For identification
 - Paternity testing
 - Forensic testing
 - Ancestry testing

Overview of selected technologies

- Chorionic Villus Sampling and Amniocentesis



- Preimplantation Genetic Diagnosis with in vitro fertilization (PGD)



Coming to a clinic near you...

Genome-Wide Fetal Aneuploidy Detection by Maternal Plasma DNA Sequencing

*Diana W. Bianchi, MD, Lawrence D. Platt, MD, James D. Goldberg, MD, Alfred Z. Abuhamad, MD, Amy J. Sehntert, MD, and Richard P. Rava, PhD, on behalf of the MatErnal BLood IS Source to Accurately diagnose fetal aneuploidy (MELISSA) Study Group**

ARTICLE

doi:10.1038/nature11251

Non-invasive prenatal measurement of the fetal genome

H. Christina Fan^{1†*}, Wei Gu^{1*}, Jianbin Wang¹, Yair J. Blumenfeld², Yasser Y. El-Sayed² & Stephen R. Quake^{1,3,4}



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BIOETHICS

One Step Closer to Designer Babies

New Noninvasive Prenatal Genetic Testing Could Change Human Pregnancy Forever



SOURCE: [Flickr/o5com](https://www.flickr.com/photos/o5com/)

A new approach to testing the genes of early-stage fetuses could radically alter the experience of pregnancy and parenting from as early as five weeks, leading to a potentially dangerous moral quandary.

Thriving

Boston Children's Hospital's pediatric health blog

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SEARCH

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Will babies with Down syndrome slowly disappear?

by BOSTON CHILDREN'S HOSPITAL STAFF on JANUARY 20, 2011

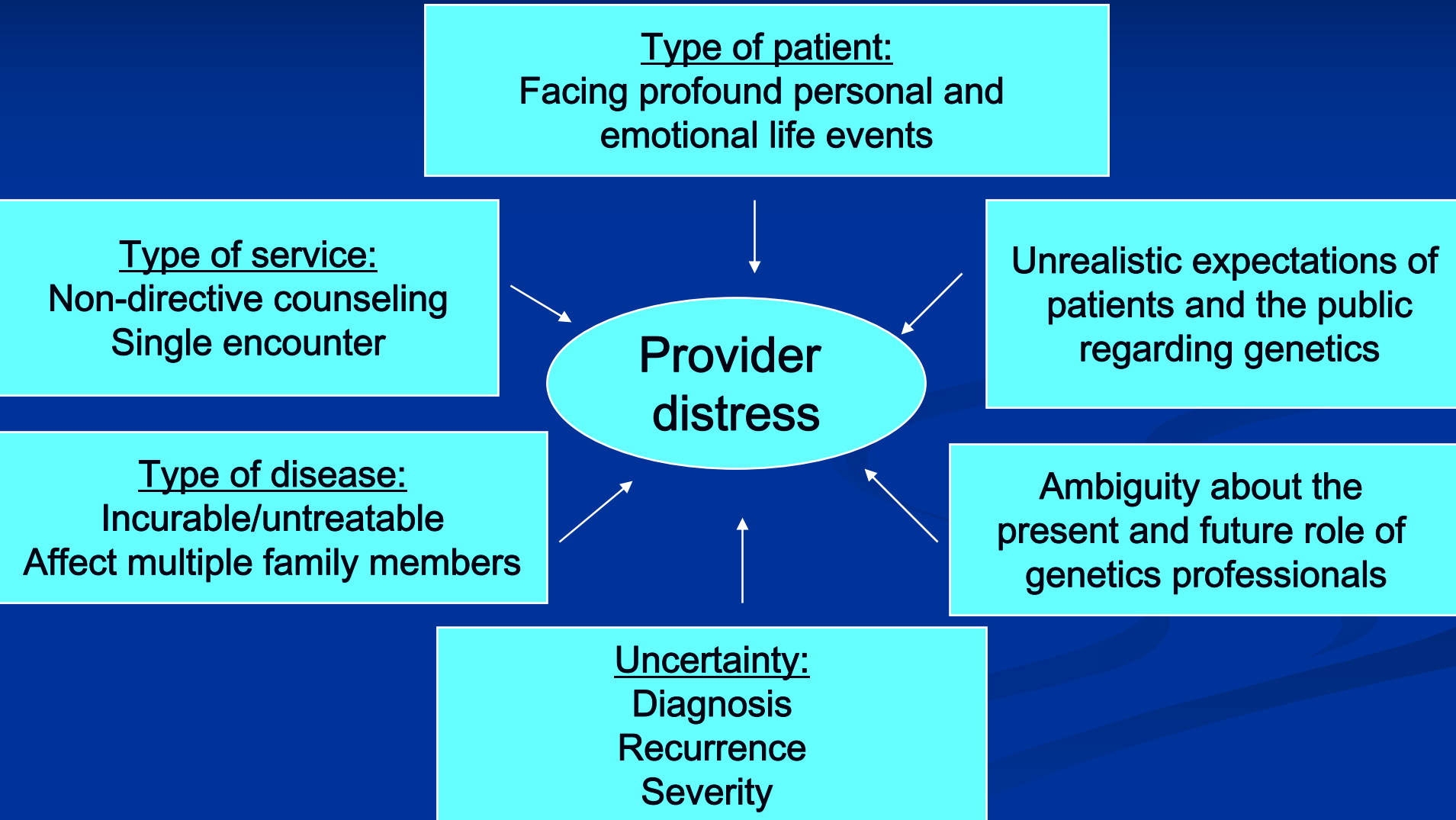
Written by [Brian Skotko, MD, MPP](#)

Children's Hospital Boston Clinical Genetics Fellow, Down Syndrome Program

Genesis of our distress and suffering grant

- New technologies raising moral and ethical quandaries for geneticists
- Evidence that genetic counselors are leaving the profession
- My own distress
- Gaps in research and training
- My colleague's encouragement

Possible sources of distress for genetic service providers



Project aims

- To examine the nature, extent, sources and consequences of distress experienced by genetic service providers
- To develop recommendations regarding interventions to reduce or manage distress

Methodology

- **Focus groups to explore the nature of distress**
 - Clinical geneticists
 - Genetic counselors
 - Nurses in genetics
- **Converted themes into survey items**
- **Surveyed 480 genetic service providers (160 of each type)**
- **Conducted interviews with a subset of survey respondents (n=54)**

Focus Groups

What clinical experiences
“keep you up at night”?

“ I was pregnant at 37, and it was assumed in the division that I would have an amniocentesis, and I chose not to do so, and I was really ostracized for that because they asked me the reason, and I was honest and said, “I know I’m at risk for was Down's, and I wouldn't abort a Down's”...my boss decided that from that point forward, I couldn't see prenatal patients because I was obviously too prejudiced about my thinking to be able to be nondirective...”

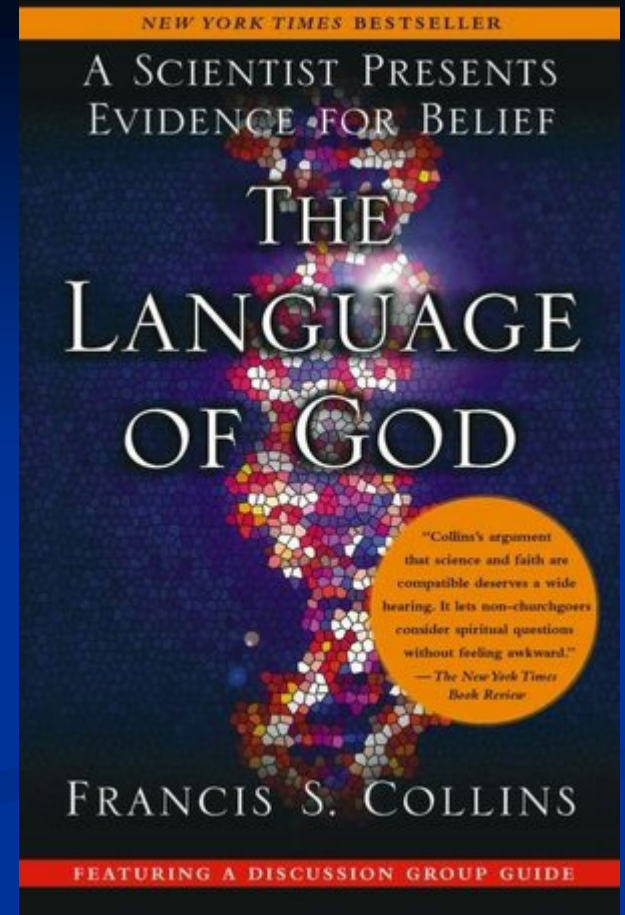
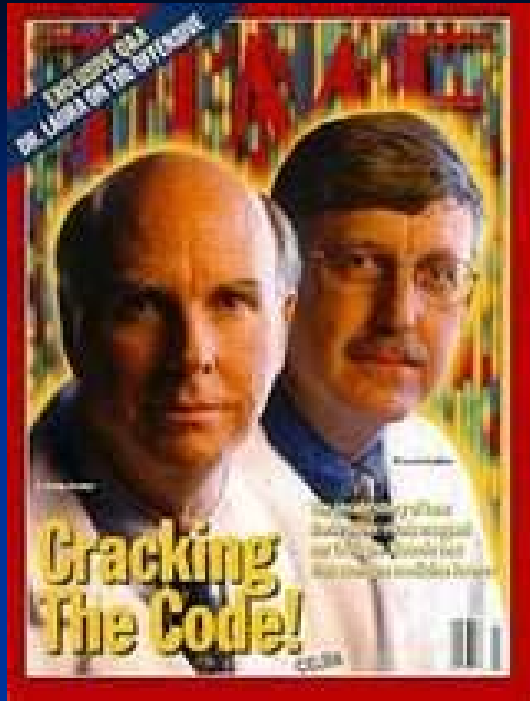
“...at this meeting a few years ago, a presentation was given by someone I respect a lot, but in my opinion, the talk made fun of religion, and religious beliefs... It gave me the sense that in this setting, and with these professionals, religion is undermined, and sort of made fun of. And that is something near and dear to my heart...All across the board it's like, if you are smart and scientifically inclined, then there is no place for faith...”

Literature on Religion & Genetics

- Theological debates re: advances in reproductive and genetic technologies
- Public attitudes about whether genetic interventions = “playing God”
- Patients’ reliance on religion/faith/ prayer to
 - explain the development of disease
 - cope with adverse outcomes
 - guide decision-making regarding tests and treatment

Literature on Genetic Service Providers and Religion

- Compared to the general US population, Genetic counselors are less likely to:
 - Believe in god
 - Attend religious services
 - Pray
- Genetic counselors who are less religious or spiritual are less likely to engage clients in discussion of religious issues



“The God of the Bible is also the God of the genome. He can be worshiped in the cathedral or in the laboratory”.

(Collins 2006)

Survey Development

- Included 38 items related to possible sources of distress
- 4 of the items related to “personal values conflicts”
 - Feels conflicted about disclosing personal beliefs to colleagues
 - Has difficulty reconciling one’s own faith with being a genetics professional
 - Feels the need to hide one’s own faith from colleagues
 - Feels like one’s professional behavior is not consistent with one’s personal values

Survey Results

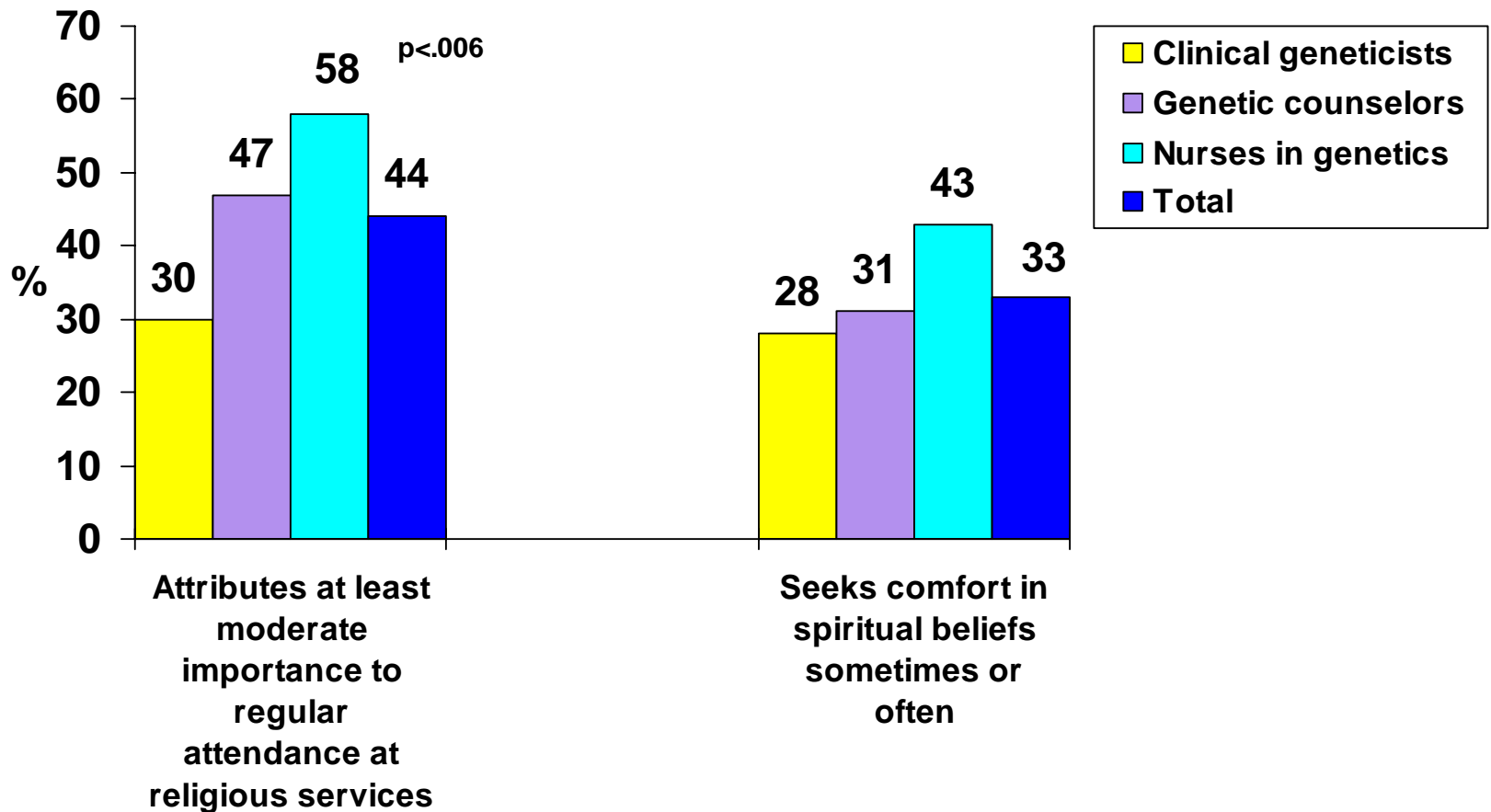
Survey Respondents: Demographics by Discipline (N=214; 61% estimated response rate)

Characteristic	Clinical Geneticists n=72	Genetic Counselors n=82	Genetic Nurses n=60
Age: mean years	52	35	49
Marital Status: % Married	89	72	83
Gender: % Female	46	84	97
Racial Background: % White, non-Hispanic	90	84	88

Respondents: Practice Characteristics By Discipline

Characteristic	Clinical Geneticists	Genetic Counselors	Genetic Nurses
Mean % Time Seeing Patients	56	72	57
Years in Practice, %			
< 5	6	38	7
5-10	11	27	14
>10	83	35	79
Type of Practice Setting			
% Academic medical center	67	50	43

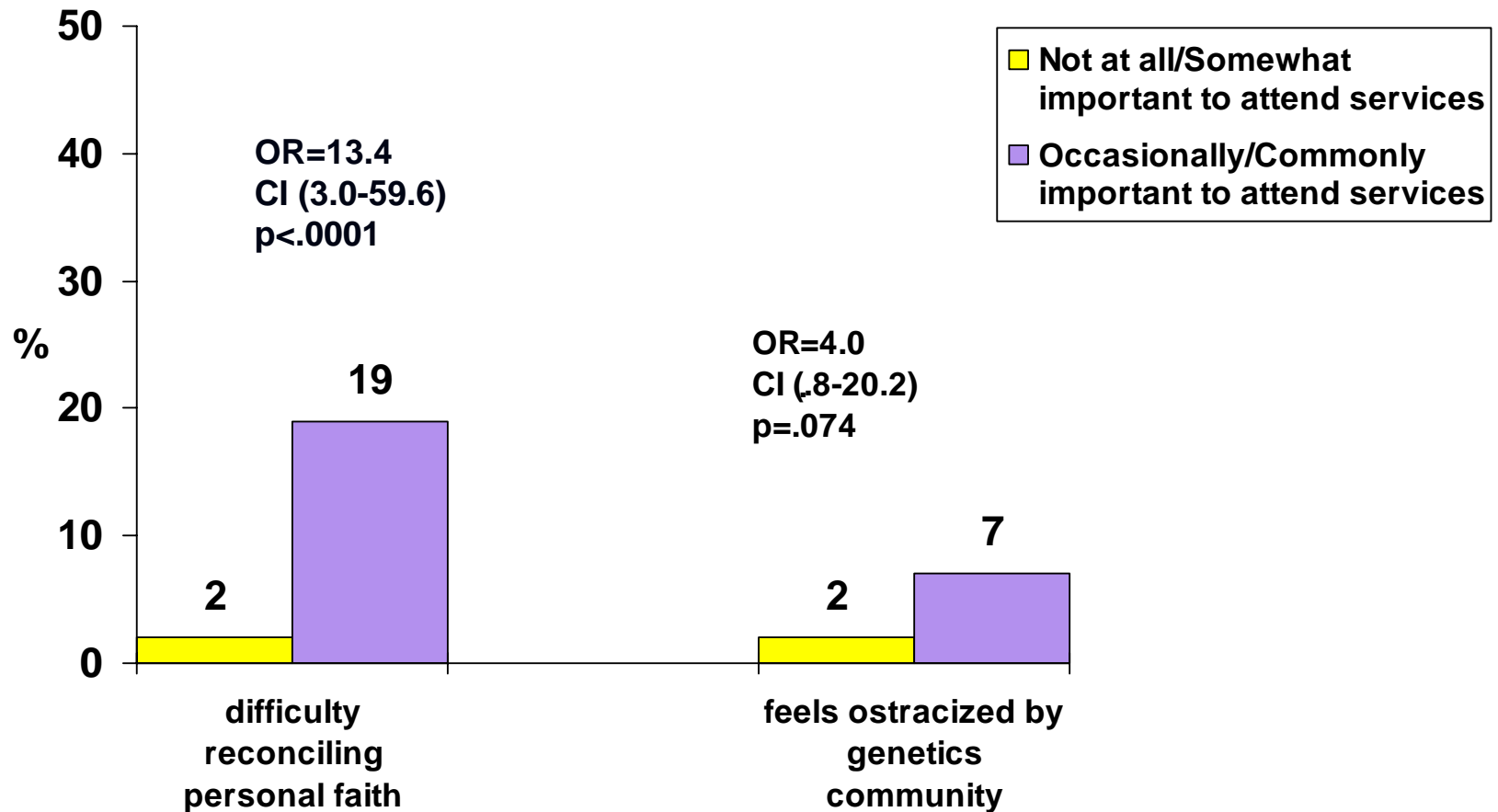
“Religiosity” by Discipline



Genetic Counselors' experiences and impact of personal faith (N=81)

	# responding occasionally/ commonly	%
Feels conflicted about disclosing personal beliefs to colleagues	25	31
Has difficulty reconciling one's own faith with being a genetics professional	15	19
Feels the need to hide one's own faith from colleagues	11	14
Feels like one's professional behavior is not consistent with one's personal values	9	11

% Experiencing Personal Value Conflicts by "Religiosity"



Correlations Between “Religiosity” and Burnout

Burnout	Importance attributed to regular attendance at religious services		Seeks comfort in religious and spiritual beliefs	
	r ²	p	r ²	p
TOTAL	-.19	.005	-.13	.061
Emotional exhaustion*	-.17	.012	-.072	.296
Depersonalization**	-.19	.006	-.16	.02
Personal accomplishment***	.12	.096	.13	.06

Interviews

Interview participants*

	Clinical Geneticists	Genetic Counselors	Nurses	Total
Male	9	5	0	14
Female	6	20	14	40
Total	15	25	14	54

* There were no systematic differences in demographic or practice characteristics between those who chose to be interviewed and those who did not.

Those who are not distressed by their religious beliefs

- Can keep their two worlds separate

“... More than anything, with what I do my faith is reinforced... I think that they just work together for me. I do not feel particularly challenged. Someone deciding counter to what I may believe, it really does not distress me too much.”

Those who are not distressed by their religious beliefs

■ Find a way to merge their two worlds

“I consider myself to be a religious person...but I had not thought to pray for my patients. It just all of a sudden came to me. They probably need it more than I do. When I am praying for them I am praying for me to be able to give them good guidance and also for them to be able to make the decision that is right for them...I am praying that I will not be pushing them in any direction and not be putting my values on their decision.”

“sometimes when you develop these relationships with these people, you get a hint about how their faith sustains them. People who have a faith, I think, are comfortable if you mention their faith and what kind of things they can do to help them through this thing...People who say they have no beliefs, I don't believe, because sometimes when I would say to them I will pray for you, they would say “Thank you.”

Those who are not distressed by their religious beliefs

- Benefit from using faith in their work

“...praying for this patient that you dislike helps you develop empathy for them... You are thinking about what they are going through instead of just drawing their family tree and working up what they are likely to test positive for.”

“I usually pray. I find that when something is plaguing me, that the best coping mechanism that I can use is to give it up and let God take it... If I get busy, I notice that I am not [praying] and I notice I am not as good at what I do.”

Those who are distressed by their religious beliefs

- Are reluctant to disclose their personal faith to colleagues

“...I do not say, “I do not believe in termination” because I am worried about people judging me for feeling like it would be immoral for me to have a pregnancy termination. I do not really express my views.”

“I have told people in the past that even if I got pregnant, I might not do prenatal testing and that was just a complete shock...you could tell by the look on their face that they thought that was crazy.... Of course as nondirective counselors, nobody would say anything about your views being wrong.”

Those who are distressed by their religious beliefs

- Feel conflicted when seeing prenatal patients

“...what bothers me the most is that sometimes I feel like when patients come in for genetic counseling, that they are very defensive in the fact that they would not terminate a pregnancy. I have had a couple of patients that were conservative in their religion or whatever and they felt very uncomfortable even telling me that they wouldn't terminate a pregnancy...because they think our whole profession opposes religiously-based decisions”

Those who are distressed by their religious beliefs

- Feel that scientific advances and professional obligations require them to set aside their personal faith

“I was born and raised Catholic.. Scientists do find things based on embryonic stem cells and people do get better...but the other side is that we are killing life. For me an embryo is a life...I cannot imagine doing that, but I want to be able to help people that are alive now...My bosses would understand if I removed myself, but I want to stay involved in the science and sometimes I worry that [its] at my spiritual peril. ...I just have to shut that button down”.

Those who are distressed by their religious beliefs

- Judge patients who rely on God to cope with adverse outcomes

“...when situations come up that are bad for our patients,..I tend to be frustrated by people who say, this is God’s will or we are going to pray or this was meant to be, that kind of stuff,..I respect it on an interactive level with them, but I hate it! I think it is wrong...”

Conclusions

- **A significant minority of genetic service providers**
 - are religiously observant
 - rely on their religious values to cope with distress
- **Some experience difficulty**
 - reconciling their religious beliefs with the expectations of their profession
 - sharing their beliefs with their colleagues and patients
- **Such personal value conflicts are more prevalent in the prenatal setting and among genetic counselors**
- **Providers who are more “observant” are less likely to experience distress from patient care and are at lower risk for burnout**

Recommendations

- **Efforts should be made to prevent or reduce the secrecy surrounding personal faith and religion among genetics professionals.**
- **Greater openness and tolerance of religious differences between genetics professionals will improve intercollegial relationships and serve as models for culturally competent patient care.**

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Publications relating to this project:

Geller G, Bernhardt BA, Carrese J, Rushton CH, Kolodner K: What do clinicians derive from partnering with patients?: A reliable and valid measure of "personal meaning in patient care" *Patient Educ Couns* 72: 293-300, 2008.

Bernhardt BA, Rushton CH, Carrese J, Pyeritz RE, Kolodner K, Geller G: Distress and burnout among genetic service providers. *Genet Med* 11(7): 527-535, 2009.

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