

Personalizing Genomic Information

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Overview

- Current genetic counseling practice
 - Approaches
 - Nondirectiveness
 - Process
- Anticipated similarities and differences in genetic counseling for genomic profiles
- Research questions



Genetic counseling: Session structure

- Contracting/agenda setting
- Information gathering (e.g., family history, medical history, psychosocial)
- Information sharing (education)
 - Basic genetics, natural history of disorder, genetic testing, management, risk to other family members
- Risk assessment and risk communication
- Informed decision making
- Plan for follow up



Service Delivery

- Primarily face-to-face individual/family counseling
 - 30-60 minutes
 - Generally some physician involvement
- Telemedicine
- Telephone genetic counseling
- Group genetic counseling
 - Lecture format
 - May be followed by individual counseling
- Passive forms of education: brochures and websites



Settings and Indications

- Reproductive genetics, IVF
 - Risk assessment, risk communication, testing options
- Pediatric genetics and metabolic disease, general adult genetics
 - Diagnostic evaluation, testing, management
- Cancer, cardiovascular, neurogenetics, psychiatric genetics
 - Risk assessment, genetic testing, result interpretation, risk reducing management strategies when applicable

Genetic Counseling Approaches: Teaching Model*

- Goal: Educated counselees
- Perception: clients come for information
- Assumption: if informed, clients can make own decisions
- Assumptions about human behavior and psychology simplified and minimized. Cognitive and rational processes emphasized
- Counseling task is to provide information as impartially as possible
- Education is an end itself
- Relationship with patient based on authority rather than mutuality

*Kessler S (1997). *Journal of Genetic Counseling* 6:287-295.



Counseling Model*

- Goals
 - To understand the other person
 - To bolster their inner sense of competence
 - To promote greater sense of control over their lives
 - Relieve psychological distress, if possible
 - To support and possibly raise their self-esteem
 - To help them find solutions to specific problems
- Perception: Clients come for counseling for complex reasons (e.g., information, validation, support, anxiety reduction)
- Complex assumptions about human behavior and psychology that are addressed



Counseling Model

- Counseling tasks
 - Assessment of strengths, limits, needs, values, and decision trends
 - Requires range of counseling skills to achieve goals
 - Requires individualized counseling style to meet needs and agendas; flexibility
 - Requires counselor to attend and take care of self
- Education is a means to achieve above goals
- Relationship aims for mutuality

**Kessler S (1997). Journal of Genetic Counseling 6:287-295.*



Teaching versus Counseling

- Leaders in the genetic counseling field advocate a combination of both
- Ideally, providers need to have the flexibility to apply the appropriate model for any given client situation-**personalized** approach
- Key goal of genetic counseling: facilitating decision making. Either model better suited to meet this goal?
 - Is information sufficient to drive informed decision making?



Factors the Drive Approach Selection

Counselor/clinician factors

- Training
- Perspective on value of education versus counseling
- Clinic logistical factors
 - Time allotted
 - Role on team
 - Importance of other components of visit

Client factors

- Stated needs
- Indication for genetic counseling
- Decision-making style
- Complexity of decisions
- Personal/family experience
- Distance between perceived and actual risk
- Perceived control
- Health beliefs and culture
- Educational background
- Emotional reaction
- Support resources



Role of Nondirectiveness

- Historically, guiding principle of genetic counseling
- Purpose: To promote patient autonomy and distance genetic counseling from eugenics- informed, autonomous decision making
- Problems:
 - Varying definitions
 - Unattainable goal?
 - Limits full use of counseling techniques and engagement
 - Questionable efficacy with regard to informed decisions
 - May be less applicable to certain counseling situations
- Practice considerations: With experience, more directive



Nondirectiveness: Workshop Report*

- Role must be clarified
 - May still serve as a component of clinical practice in terms of supporting and respecting clients beliefs, values, and traditions
 - Not a theory of practice
- There is a need for a flexible approach to genetic counseling with varying adherence to nondirectiveness based on client/family needs, values, clinical circumstances and desired counseling outcomes.

Weil J et al (2006). Journal of Genetic Counseling 15(2): 85-93.



Current Practice-Process

- Limited but growing number of studies have evaluated the content & process
- Analysis of 18 process studies*
 - Providers speak more than clients
 - Large proportion of communication is biomedical rather than psychosocial
 - Teaching model is widely implemented
 - Higher level of counselor facilitation of understanding and empathic responses, lower verbal dominance associated with more positive client outcomes

* Meiser B et al (2008): *Journal of Genetic Counseling* 17:434-451



Limits of translating experience in current practice to genomic profiles

- ❑ Growing but still limited body of research regarding process and outcomes
- ❑ Alternative models of delivery relatively new and not fully evaluated in terms of communicating traditional genetic information
- ❑ Impact of internet, social networking and web-based resources not fully appreciated
- ❑ Much of existing research evaluated communication by genetics professionals (geneticists, genetic counselors, genetic nurses)



Genomic versus Genetic Information-Similarities

- By virtue of being genetic
 - Familial information
 - Stigmatizing in certain cultures
- Can be associated with a degree of uncertainty
- Will be those who seek out information for health promotion

Genomic Information Versus Genetic Information-Differences

Genomic Profiles

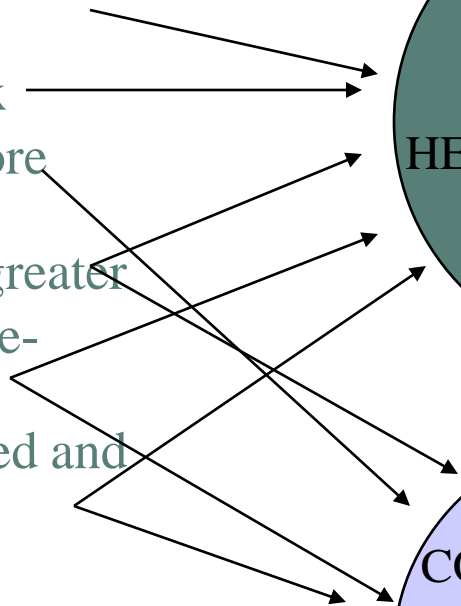
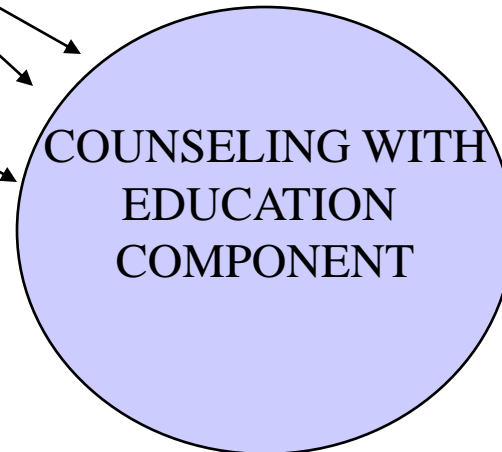
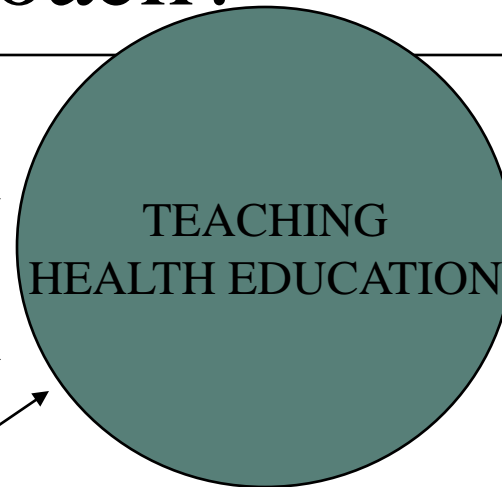
- Volume of information
- May not be associated with family history
 - No context
 - Requires belief that information is valid
- Available to everyone
- Significant proportion potentially more actionable with regard to health promotion

Genetic Information

- Tends to be targeted
- In context of chronic disease, often associated with family history
- Uptake for some services (e.g., cancer genetics) more likely among those with higher SES and education levels
- Only subset is actionable re: health promotion

What does this mean in terms of genetic counseling approach?

- ❑ Stated needs- may be less clear
- ❑ Indication for genetic counseling- more complex
- ❑ Decision-making style-more varied
- ❑ Complexity of decisions-greater
- ❑ Personal/family experience-variable
- ❑ Distance between perceived and actual risk- could be great
- ❑ Perceived control-varied
- ❑ Health beliefs and culture-more varied
- ❑ Educational background-varied
- ❑ Emotional reaction-varied
- ❑ Support resources-varied





Assessing Value: Research Questions

- Desired outcomes of genomic/personalized medicine- value
 - Informed decision making
 - Health promoting behaviors
 - Perceived control and low distress
- Research questions
 - What is the most effective way to communicate this volume of information that promotes these values?
 - How do you identify which approaches are most effective for individual clients? How does this have an impact on choosing service delivery models?
 - Currently genetic counseling focuses on informed decisions and health promotion through facilitating understanding. Is understanding enough or are there other barriers to action?
 - Is there value in incorporating motivational interviewing techniques to genetic counseling?



Summary

- Given client variables, effective communication of genomic profiles seems best suited for a blended teaching/counseling approach
- As part of clinical utility studies, need to develop a body of literature that evaluates how to maximize perceived value and intent to act and then develop evidence-based counseling strategies.