Using Genomic Evidence for Patient Decision Making: A Nursing Perspective.

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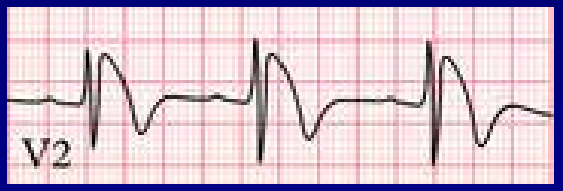
The Nurse Practitioner's Role in Genetics

- Taking a comprehensive 3-generation family pedigrees
- Recognizing signs and symptoms of genetic conditions
- Ordering diagnostic and genetic tests
- Providing counseling and education
- Support to patients and families
- Follow-up monitoring of treatment

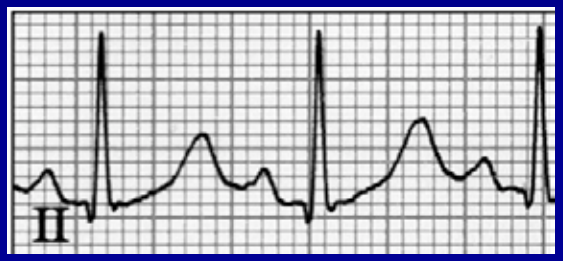
Clinical Finding Examples

Targeted Panel

Hypertrophic Cardiomyopathy Panel



Brugada Panel



LQTS Panel

What comes first?
Targeted Panels or Whole-Exome?

Targeted Panel ⟶ Whole Exome

Examples:

- HCM (20 genes)
- Brugada (7 genes)
- LQTS (12 genes)

- Next generation sequencing
- (Microarray - 10% will have a large deletion/duplication)

Shared Decision Model for Cardiac Care

- A young female suffers an out of hospital cardiac arrest.
- No cardiac or family history
- Normal ECG, echo, exercise test, MRI, etc.
- SCA panel was ordered (10 gene panel) which was negative.
- Shared decision-making model by patient and providers to order a whole exome.
- Whole exome revealed a rare variant.

Reimbursement Considerations

Targeted Cardiac Panels: $3,375 per panel

Whole Exome Sequencing (Trio: Parents + Proband): $9,000

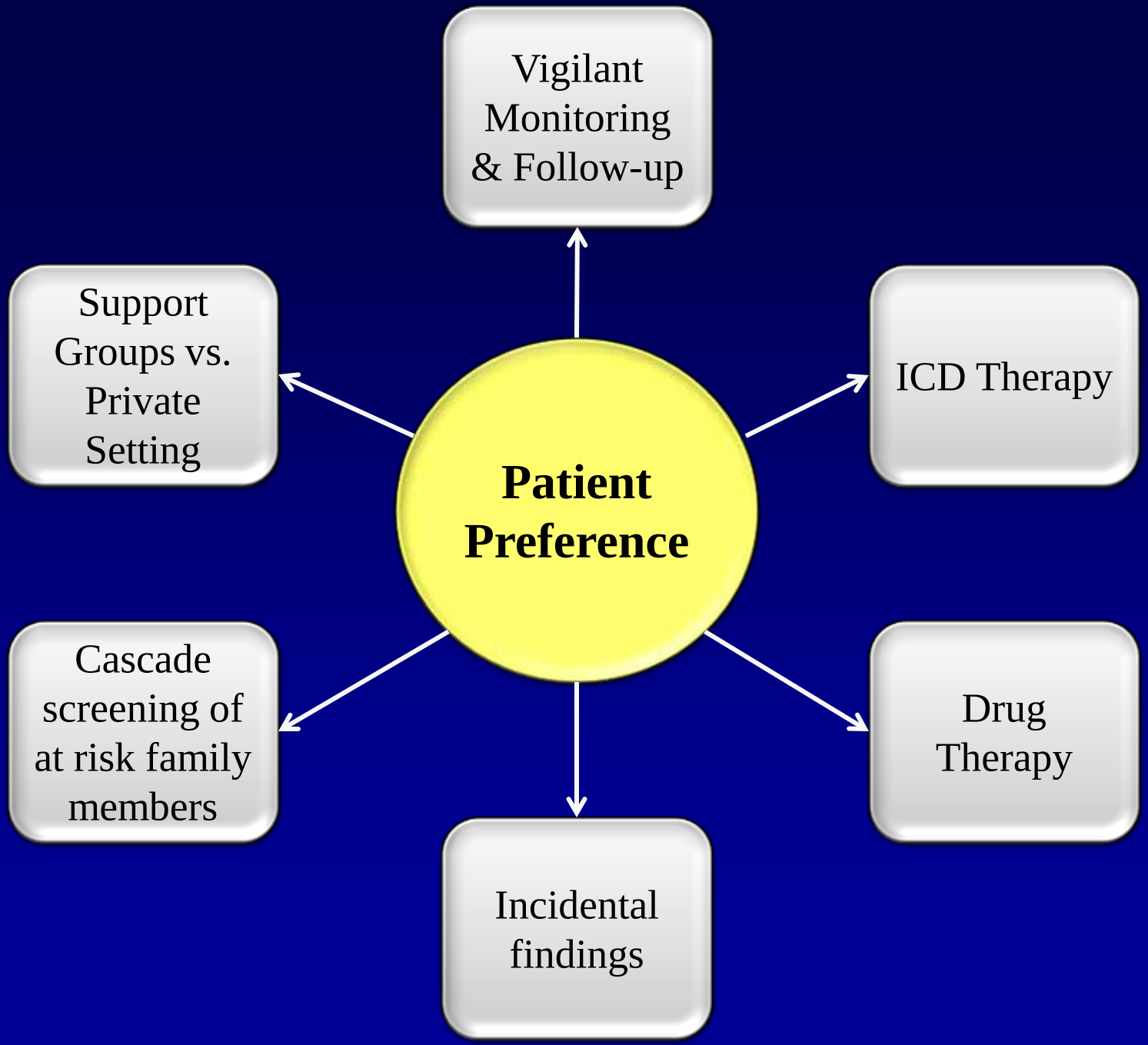
Whole Exome Sequencing (Proband): $5,000

Whole Exome Sequencing for Additional Family Members: $2,500

Uninsured: Case-by-case/Payment Plan Option

The Real World Setting

- Serving the diverse population of NYC
- Various levels of health literacy, socioeconomic backgrounds
- Access to care and comprehension
- Patients tend to feel overwhelmed saturated by information in about 30 minutes
- Nurses play a key role in helping individuals understand the complex language
- Sharing the real details-- the nurse practitioner is first in the room & the last to leave


Vigilant Monitoring & Follow-up
Support Groups vs. Private Setting
ICD Therapy
Patient Preference
Cascade screening of at risk family members
Drug Therapy
Incidental findings

How does a cardiac diagnosis impact patients?

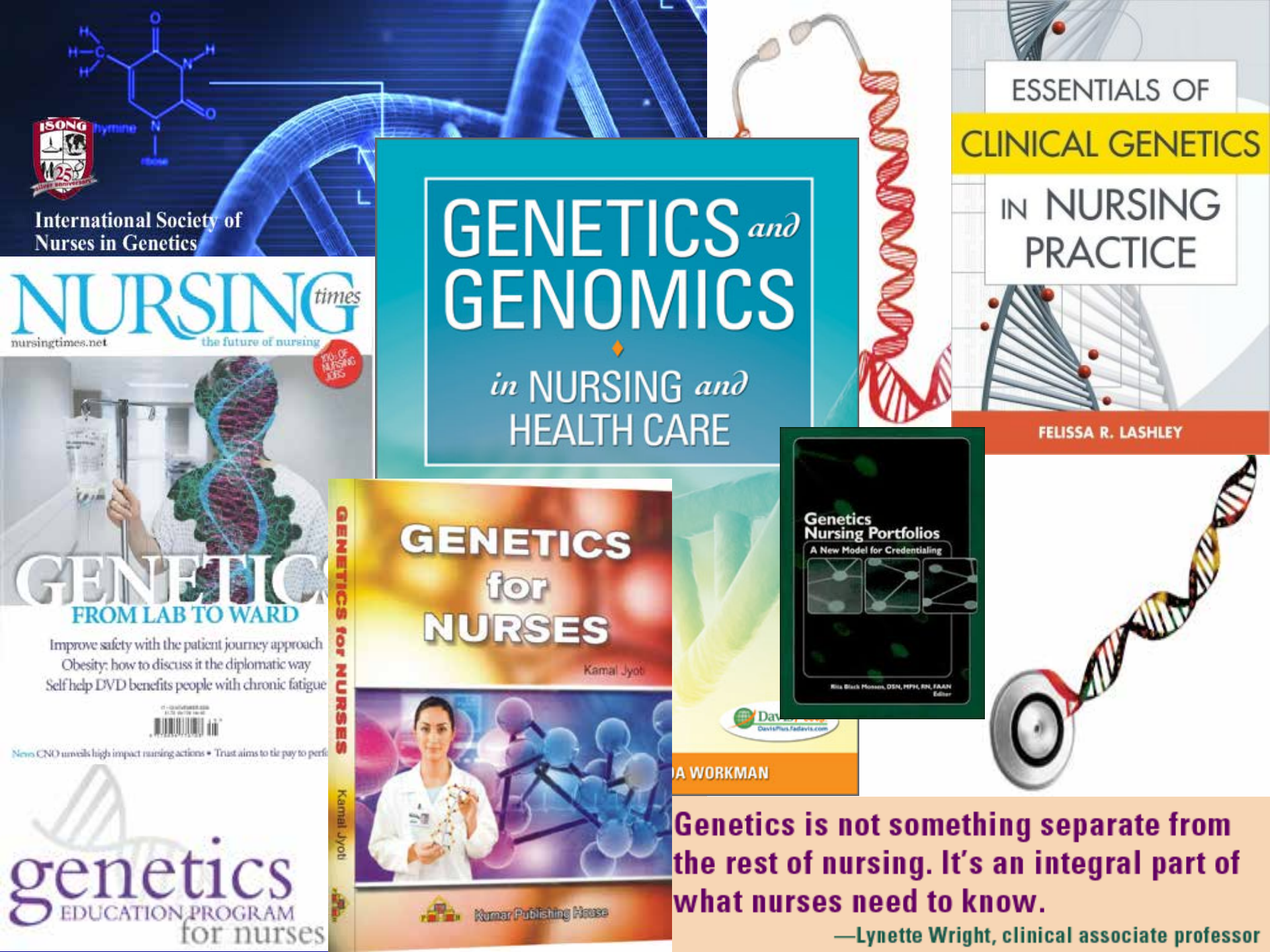
Table 1. Demographics	
Variables	
Mean Age (years ± SD)	42 ± 11
Marital status (Married)	23%
Education Status	
High School	82%
Some College	10%
Income less than 55,000/per year	96%
Insurance through Medicaid	98%
Family history of SCD under the age of 50 years	33%

N=31
Hispanic Participants

Table 2. Quantitative Results	
SF-36 Questionnaire	Mean ± SD
Physical Component Summary	46.2 ± 6.6
Mental Component Summary	59.9 ± 5.3

Table 3. Identified Themes	
Themes from Semi-Structured Interview	Patients Reported
Fear of Sudden Death	89%
Guilt of Passing on a Genetic Mutation to their children	77%
Fear of ICD firing	73%

Robert Wood Johnson Foundation Nurse Faculty Scholar Awards given to Kathleen T. Hickey (Award #: 66515, 9/1/2009-2/28/13).



Genetics is not something separate from the rest of nursing. It's an integral part of what nurses need to know.

—Lynette Wright, clinical associate professor