Objectives

- introduce the Department of Genetics and Regional Genetic Services
- appreciate the impact of genetic disease on primary care
- identify some recent advances in genomic medicine
- who and when to refer to genetics
- preview the genetics symposium tomorrow

Question #1

What is your clinical practice?

1) Internal medicine
2) Internal medicine subspecialty
3) Family medicine
4) Hospitalist
5) Other
Have you referred at least one patient to genetics in the past three months?

1) Yes
2) No

Can you name or recognize your local geneticist or a genetic counselor?

1) Yes
2) No
Who We Are

- 9 clinical geneticists (MD)
- 31 genetic counselors (MS)
- screening programs staff
- admin/support staff

What We Do

- preconceptional & prenatal counseling
- genetic screening: prenatal & newborn
- clinical genetic services for patients of all ages
- cytogenetic, biochemical & molecular diagnostic testing
- educate members & health care providers on genetic issues

What Are Our Tools?

- patient’s medical history
- family medical history (“it’s the pedigree, stupid!”)
- up-to-date web resources
- cytogenetic tests
- biochemical tests
- molecular tests
- effective counseling
Sites We Serve

The Changing Medical Marketplace

- increased competition
- all vendors hawking their own
  “KP Promise”: convenience, affordability, quality, great service
- "the new normal"
- innovation
- process improvement
- replicate success

Genetics Telemedicine

Implemented 2011 in Antelope Valley & Kern County
- patients offered in-person or telemedicine options

Real-time Clinical Genetics Encounters
- consult and flu visit types in Cadence
- preconception, prenatal & cancer genetics
- co-pays apply
Innovations in Genomic Medicine

Cell-free fetal DNA

Genetic Counseling and Testing
Where Can We Improve?

- accurately diagnose all members with genetic disease (refer)
- improve management of members with genetic disease (integrate)
- identify members at risk for genetic disease (family hx)
- educate members and MDs about genetic diseases
- decrease morbidity and mortality

When to Refer to Genetics

- OB: AMA, >3 miscarriages, (+) family hx
- Neo: birth defects, dysmorphic, abnormal newborn screens
- Peds: dysmorphic, developmental delay, short stature, (+) family hx
- IntMed/FamMed: affected with or (+) family hx of genetic disease
• Who’s yo’ daddy?
• What’s my ancestry?
• molecular confirmation of clinical dx
• isolated cancer >50 yr old (neg fam hx)
Translating Genes into Healthcare

The Kaleidoscope of Genetic Testing

What Can Genetic Counseling Do for You & Your Patients: Bridging the Gap

Tell me, how does this mutation make you feel?
Keynote: The Future Is Genomic Medicine
Wayne W Grody, MD, PhD
UCLA School of Medicine
President, American College of Medical Genetics and Genomics

“The transition from traditional targeted gene testing to genome-wide analysis constitutes a genuine sea change in medicine, offering vastly enhanced diagnostic power along with unprecedented challenges in test interpretation and reporting.”

Hereditary Breast Cancer: The Power of Defining Risk

- Breast cancer (often <50yr) (40–85%)
- Contralateral breast cancer (40–60%)
- Ovarian cancer (10–40%)

Scoping Out Hereditary Colorectal Cancer
Common Adult Genetic Disorders:
A Primer for Primary Care

Cardiovascular Genetics:
Getting to the Heart of the Matter

Genetics in Your Clinical Practice
Presented by the Department of Genetics and Regional Genetic Services, Kaiser Permanente Southern California
September 22, 2012