Genetic Counseling

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Overview

- Genetics – a Growing Field
- Genetic Counseling
- Applying to Programs
- Northwestern University
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There are more than 50 well described genetic cancer susceptibility syndromes

Genetic counseling and testing shortly after cancer diagnosis can help guide patients’ surgical and treatment decisions.
34% reduction in chemotherapy use would occur if women with breast cancer receive a genetic test prior to treatment.

17,000 strokes could be prevented each year if a genetic test is used to properly dose the blood thinner warfarin.
Ten neurogenetics tests in 1995, **several hundred today** (Charcot-Marie-Tooth neuropathy, hereditary ataxia, spastic paraplegia, muscular dystrophy and early-onset dementia)


Lifetime risk of developing Alzheimer disease (AD) is 10-12%. **Few clinicians are prepared to address the genetic risks of Alzheimer disease** with their patients

ACOG currently recommends that Down syndrome screening and invasive testing be offered to all women, regardless of age.

3-5% of all babies born will be affected with an abnormality, such as a birth defect, intellectual disability of genetic syndrome.

ART (Assistive Reproductive Technology)/IVF conceived pregnancies becoming more and more common
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Why Genetic Counseling?

- 13 million Americans have a genetic disorder
- 10% of chronic diseases, such as heart conditions, cancer, diabetes, and arthritis, have a significant genetic component
- 31% of the time, physicians misinterpret results of genetic testing (NEJM 1997. 336(12):823-7)
Genetic Counseling

What does a **genetic counselor** do?

- **Traditional role**: discuss personal/family health history
- Determine level of risk for certain diseases

- **If family or personal history puts you at a high risk:**
  - Discuss the pros and cons of testing
  - Discuss implications of testing
Genetic Counseling

What does a **genetic counselor** do?

- **Nontraditional roles:** act as a consultant
  - Liaison between lab and clinician
  - Medical science liaison (Myriad genetics, GeneDx)
  - Telehealth (SIUSOM, carrier screening companies)
Genetic Counseling

Where do genetic counselors work?

- Hospitals/private clinics: cancer, prenatal, peds, adult, specialty
- IVF clinics, Private practice
- Genetic testing laboratories
- State Department of Public Health
- Health insurance companies
- Remote positions-telephone consulting
- Research settings
Where have I worked?

- **Private laboratory** (NxGen MDx)
- **University clinics** (SIUSOM, UICOMP)
- Illinois Department of Public Health – **Newborn Screening Program**
- **Blog writer** – ataxic CP, radiation
UICOMP Genetics Clinic

- **Common reasons for referral:**
  - Failure to thrive
  - Dysmorphic features
  - Developmental Delays/Autism
  - Family history of known genetic condition
  - Family or personal history of cancer
  - Abnormal prenatal ultrasound or screening
UICOMP Genetics Clinic

- **During the appointment (peds):**
  - Obtain family history
  - Obtain medical history
  - Obtain developmental history
  - Physical evaluation
  - Obtain consent for video/photos
  - Review genetic testing options
    - Insurance coverage, cost of testing
  - Recommend referrals
Our Genome

Genetic information within the cells in our bodies

- Chromosomes - set of 46 books
- Genes - pages
- DNA - letter code

Chromosomes (Karyotype)
Chromosomes

= Set of books/encyclopedia

46 total
• 23 from mother
• 23 from father

• Autosomes (22 pairs)
• Sex chromosomes (X, Y)
46,XX Female Karyotype
46,XY Male Karyotype
Genes

= pages in the books

Genes are instructions for making everything the body needs, they tell our body how to grow and function
DNA

= words on the pages
Dominant disorders are caused by mutations of spelling errors in one copy of a gene.
Achondroplasia

Hereditary Cancer Syndromes

Majority of cancer-sporadic (environment/lifestyle)
~15-20% familial (shared genes and environment)
~5-10% hereditary (caused by single gene mutation)

Hereditary colon cancer (HNPCC)
  • Dominant

Hereditary breast and ovarian cancer (HBOC)
  • Dominant
Recessive disorders are caused by mutations or spelling errors in both copies of a gene.

25% risk
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Graduate Programs

- **Almost 40** genetic counseling graduate programs in the US and Canada

- **Several** new programs under review

  Number of students matriculating in 2016 (including Canadian schools): 317

  No additional clinical training, residency etc. is required after completion of the degree, go directly into the job market!
Applying

- 3 letters of recommendation
- GRE scores
- Shadowing GCs
- Volunteer experience (crisis counseling, hospice, special olympics, etc.)
Applying

- Become familiar with hot topics in genetics
  https://www.cdc.gov/genomics/update/current.htm

Familiarize yourself with health care laws surrounding genetics (GINA,

Take a look at 23andme website (good intro to direct to consumer testing)

National Society of Genetic Counselors (NSGC)

Genetic Task Force of Illinois (GTFI)
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Northwestern University
Northwestern

- Located on the Downtown Chicago campus
- Over 25 clinical rotations sites across Chicago

- Internship – application due early February

http://www.cgm.northwestern.edu/education/graduate-genetic-medicine/summer-internship.html
Thanks for Listening!
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