Family History in Primary Care Current Practices

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Family History: The Gateway to the Molecular Age of Medicine

- Tracy L Trotter, MD, FAAP

- February 2006

“ All patients have genes.”

Eugene Rich, MD 2004

"A pedigree in every patient's chart."

NCHPEG [Robin Bennett, MS, CGC] 2004

THE CRANE'S FOOT

John Muir Medical Center

February 10, 2005

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pedigree, *n.*

- 15th c. French: ***pied de grue*** [crane's foot]
- "a conventional mark consisting of three curved lines, which bears a distinct resemblance to the claws of a bird. Used in denoting succession in pedigrees."
  - OED 1999

Why Primary Care Providers?

- 882 MD clinical geneticists
  - Patient care = 30% of their time
- 300,000,000 patients
  - “All patients have genes”
- 1 FTE geneticist per 1,136,363 patients

  - American Board of Medical Genetics 2003

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- Primary Care Pediatrician
  - San Ramon Valley Primary Care
  - Northern California – Suburbia
  - 7 Pediatricians
  - 2 Pediatric Nurse Practitioners
  - 5 Internists

Summer Camp

- A 10 yo patient has two syncopal episodes while attending Wake Forest University basketball camp.
  - Taken to ER – “workup normal”
  - To follow up with PCP
  - Seen in the office two weeks later

Family History

- MGM died of “heart attack” at age 51 yrs
- Mother “faints all the time...not a big deal”
- Mother’s sibling died of SIDS @ 10 weeks
- Father/older brother hx non-contributory
- Sister born at 32 weeks – NICU 4 weeks
  - Two episodes of bradycardia, hypotonia and cyanosis in first 10 days of life
  - Responded to stimulation – no CPR
  - Extensive workup = WNL

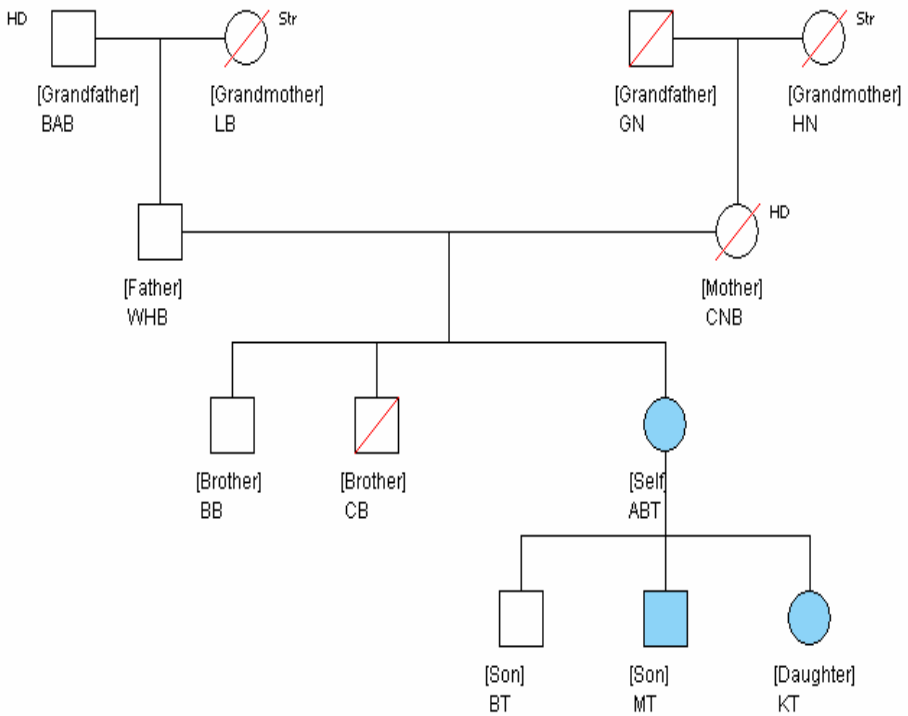
My Family Health Portrait - Drawing Report

ABT - February 10, 2006

Highlighted disease: LQTS

□ - Male Family Member
○ - Female Family Member
■● - Family Members with a History of Disease
⊠⊘ - Deceased Family Member

HD - Heart Disease
Col - Colon Cancer
Str - Stroke
BrC - Breast Cancer
Dia - Diabetes
OvC - Ovarian Cancer



ECGs for ALL

- Father and 12 yo sibling = WNL

- Mother: QTC 480 msec
- Proband: QTC 450 msec
- Sister: QTC 480 msec

LQTS

- Long QT syndrome [Romano-Ward]
- Can lead to a fatal arrhythmia
- Syncope the most common symptom
- Autosomal dominant
- Five genes [?7] have associated mutations
- Incidence 1:7000

Molecular Genetic Testing

- Mother, proband and sister all positive
  - LQTS 1 phenotype
  - Mutation in *KCNQ1* gene
- Father, brother and uncle – no mutations
- The deceased MGM [MI] and uncle [SIDS] were likely affected.

Happy Ending

- The mother, proband and sister are on beta blockers and doing well.

Pediatric Use of Family History

- Superb diagnostic tool
- Focus evaluation and testing
- Establish a pattern of heredity
- Distinguish genetic from other factors
- Identify medical screening needs

Pedigree – Patient Education Tool

- Importance of medical documentation
- Patient can see inheritance patterns
- Demonstrate variability of expression
- Explore level of understanding
- Clarify common misconceptions
  - “Only men”
  - Skips generations”

OPPORTUNITIES

- Prenatal visit [ideal]
- Newborn visits [most common]
- New patient visit
- New symptom/diagnosis
- Query from family

SCREEN

- SC Some Concerns
- R Reproduction
- E Early Disease, Death or Disability
- E Ethnicity
- N Non-Genetic

SC *Some Concerns*

- "Do you have any [some] concerns about diseases or conditions that seem to run in the family?"

R *Reproduction*

- "Have there been any problems with pregnancy, infertility, or birth defects in your family?"

E *Early Disease, Death or Disability*

- "Have any members of your family dies or become sick at an early age?"

E *Ethnicity*

- "How would you describe your ethnicity?"
- "Where were your grandparents born?"

N *Non-Genetic*

- "Are there any other risk factors or non-medical conditions that run in your family?"

Genetic Thinking in Practice

You don't have to be an expert, but you should:

- know how to take a 3 generation pedigree
- recognize basic patterns of inheritance
- recognize general genetic red flags
- familiarize yourself with available resources
- know when to refer
- be skeptical of geno-hype and pseudoscience
- understand the implications of test results (diagnostic vs. predictive; ELSI questions)

“Why pay money to chart your family tree? Go into politics and your opponents will do it for you.”

Mark Twain