

Ethical Implications of Family History Information in Pediatric Primary Care

Lainie Friedman Ross, MD, PhD

Associate Professor, Dept of Pediatrics

Associate Director, MacLean Center for Clinical
Medical Ethics, University of Chicago

To understand the ethics, we need to understand the goals.

- To identify families with a Mendelian condition (<4% of the population).
- To identify families with complex diseases which have a genetic component.
 - Research agenda: to identify the genes responsible to improve prognostication within these families.
 - Translational agenda: to provide high risk patients with specific treatments, surveillance, or lifestyle changes that may decrease morbidity or mortality.
 - Clinical agenda: to recommend early surveillance or prophylaxis or lifestyle modifications to those who are at increased risk.

HHS Launches New Family History Initiative

- Move directly to the Clinical Agenda
 - Have we really achieved the research and translational goals?
- Six Main Diseases
 - Heart Disease
 - Stroke
 - Diabetes
 - Colon Cancer
 - Breast Cancer
 - Ovarian Cancer
- No questions on data sheet to collect racial/ethnic data
- No questions about known carrier status (despite opportunities from Newborn Screening)

Pediatric Family History Initiative

- Given what we know about genetics to date, does it make sense to focus on family history information with our pediatric patients?
- Which will be more relevant to my practice?
Knowing that 3 aunts had breast cancer or that 2 cousins who live in the same building had lead poisoning?
- Which will be more relevant to my practice?
Knowing that 3 relatives had colon cancer in their 50s versus knowing that mom never completed high school, calls her children “bad”, and uses a “switch”?

To understand the ethics, we need to understand the trade-offs.

- What affects pediatric practice today? Genetic versus Non-genetic (sociocultural) family history
 - NON-GENETIC
 - Family history of violence, poverty, or illiteracy.
 - Family exposure to violence, illicit drug use, cigarettes, and alcohol.
 - Siblings or neighbors with lead poisoning.
 - Injuries
 - Obesity
 - MIXED GENETIC / NON-GENETIC PICTURE
 - School problems
 - Hearing problems
 - Asthma and allergies
 - GENETIC
 - Mendelian conditions: CF, sickle cell disease, MCAD
 - Some pediatric cancers

What affects pediatric practice tomorrow?

- As we learn more about genetics, family histories may play a more important role in pediatric practice than they do now.
- But first, we will also need to know that this information will influence health promoting behaviors.
 - Change physician behavior (e.g. surveillance, treatment, or lifestyle recommendations).
 - Change patient/family behavior (e.g., lifestyle or health promotion activities)

The Science influences the Ethics in Genetic Testing

- If no preventive services or treatments available, labeling a person as “high risk” when asymptomatic may be unethical (e.g., 4% of the population have high risk genotypes for type 1 diabetes (T1D) ; 92% of these children will not develop T1D).
- If prevention or cure is available, failing to determine risk status is unethical (e.g., retinoblastoma).
- If increased surveillance or early treatment is possible, determining risk status may or may not be beneficial (e.g., sickle cell disease vs. neuroblastoma)
- If modifications in lifestyle are possible, determining risk status may or may not be beneficial (e.g. MCAD vs. HCM).

To understand the ethics, we need to understand the challenges raised by creating a family history genogram

Accuracy of information

Privacy and Confidentiality

Duty to Share Information

Accuracy of Family History-1

- Data show that family history is more accurate for cancer than for mental health issues.
- Data show that even for cancer, family history is only 80-90% accurate.
- How do we go about verifying the accuracy of family history?
 - HIPAA rules.
 - Time-constraints

Accuracy of Family History-2

- Consider family history of stroke.
 - You need to know not only which relatives had strokes, but which relatives have high blood pressure.
 - While sentinel events are often shared (e.g., strokes), the fact that a cousin is on an ACE-inhibitor may not be considered important family information.

Privacy and Confidentiality

- Is there an obligation to share your health information with your relatives?
 - The complexities in blended families.
- How to explain what information needs to be shared (e.g., conditions with a genetic component) versus conditions that do not need to be shared (e.g., infectious diseases unless you have exposed the relative to them [e.g. TB vs. syphilis])
- Will this increase blame and shame? Stigma and discrimination?

Duty to Share Information?

- Data show that family members share medical information NOT based on genetic ties but on emotional ties. (e.g., some women share breast cancer information with mother-in-law and not with sisters depending on social relationship.
- Some families are open about family illnesses; others are not, particularly when there are concerns about how it will be used.
 - To protect family members from insurance risks.
 - To prevent family members from using the information for reproductive purposes.
 - Because of an alternative view on genetics (e.g. Chinese Australian view the paternal uncles and cousins as brothers but maternal uncles as “non-close relatives”).

To understand the ethics, we need to consider the practicalities

- Time constraints on doing a proper family history.
- Changing family structures
- Given that 13% of births are to teenage mothers, many of the health conditions may not appear in the parents or grandparents for years to decades.
 - Need to revisit family history over time. Does it make sense to start in the newborn period?
- Increasing role of children being reared by non-biological parents (e.g. adoption or the use of donated gametes, particularly eggs for older women.

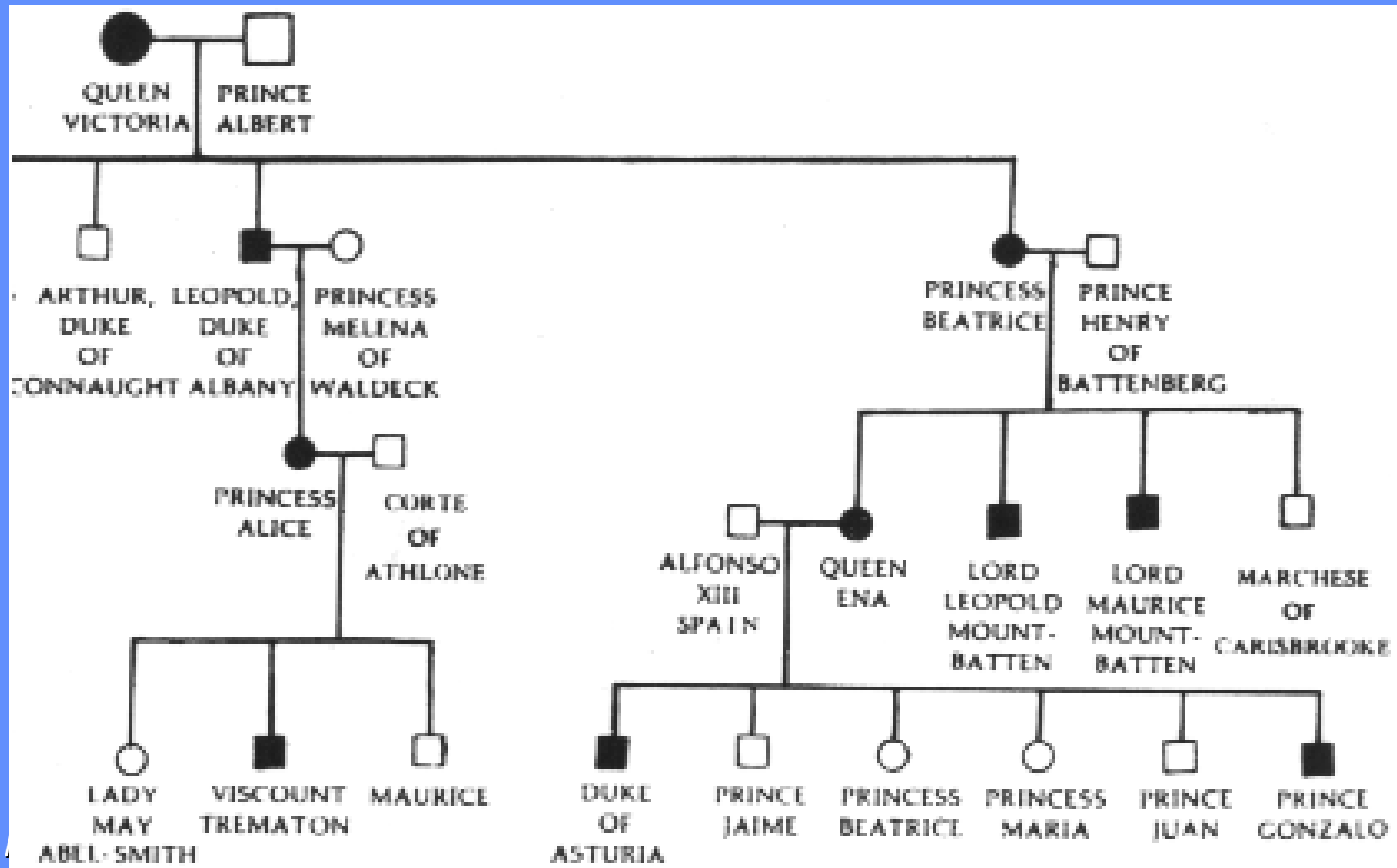
Well-Child Visit #1

- Need to review pregnancy, labor and delivery
- Need to review newborn care
- Need to confirm NBS data (blood and hearing)
- Need to provide anticipatory guidance
(including safety, developmental issues)
- Need to answer parents' questions
- Need to review office policies and procedures
- Need to obtain 3 generational family history

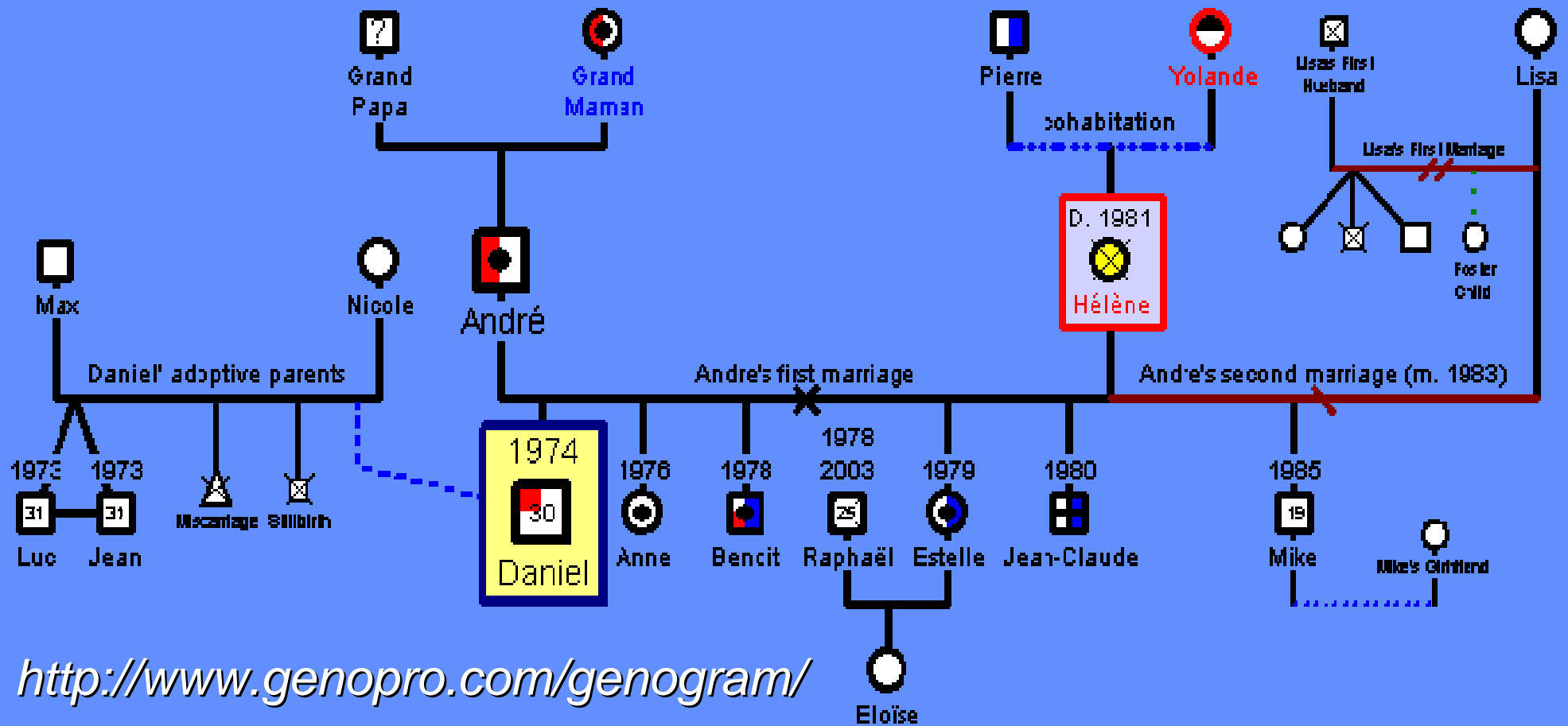
Well Child Visit #2

- Need to discuss risks and benefits of immunizations
- Need to discuss child care esp. if parent has been on maternity leave
- Need to provide anticipatory guidance (including safety, developmental issues, what is an emergency, when to introduce foods)
- Need to answer parents' questions
- Need to obtain 3 generational family history

Feasibility: A Traditional Pedigree



Feasibility: A More Modern Family



<http://www.genopro.com/genogram/>

Feasibility in Families of Teen Moms

- Although many diseases of adult-onset may not yet be present in a family when the mother is a teenager, collecting family health information may be a catalyst to learn about prior generations (beyond 3 generations) and may help families realize how important it is to communicate health information.
- It is important to encourage single teenage moms to learn more about the family history of the baby's father, particularly since many will not stay together over time.

Feasibility in Families with Older Moms

- Disclosure to the child of the genetic role of third-parties to ensure that the child knows his genetic family history \neq his social family history.
 - This will require a change from current practice of secrecy that often surrounds the use of donated gametes and “closed” adoptions.
- Increasing use of donor eggs.
 - The importance of collecting donor and donor's family health data at the time of donation and to have a system to provide for updating the info.
- Information about relinquishing parents.
 - The importance of collecting data about mother and father and their families, and to have a system in place to provide for updating the information.
- Reverse information
 - If children of donor eggs or adoptive children develop a genetic health condition, need to inform the parents as this may be relevant for their other children.

New Family History Initiative

- Should it be a research priority in pediatrics?
 - Especially important with the movement to develop pediatric biobanks.
- When will it be ready to translate the research into clinical practice?
 - When it has achieved a certain degree of clinical validity and clinical utility.
- Should it be a priority in clinical pediatrics?
 - Currently, most family history will not change the daily practice of pediatricians nor provide improved health care to children.

How to Achieve Success with the New Family History Initiative

● Education of the Public

- If the public perceives greater benefit than risk in revealing family medical information, they will be more inclined to do so with their physicians and with their relatives.
- Need educational campaign to decrease stigma and discrimination

● Education of physicians

- Physicians need to be convinced that taking a family history is as useful or more useful than other predictors of health.
- In pediatrics, physicians need to be convinced that taking a family history is more useful than time spent on anticipatory guidance.

● Systems Issues

- Need to ensure that the information will be stored confidentially.