

Utilizing the family health history (FH) to support optimal care for children



Joann Bodurtha MD MPH

Virginia Commonwealth University

A young girl with blonde hair and a young boy with brown hair are sitting at a wooden table in a library. They are looking at books on a shelf behind them. The girl is wearing a white sweater with a floral pattern, and the boy is wearing a white shirt. The background is a light blue wall with several books on a shelf. The text "So What?" is overlaid in large black font across the center of the image.

So What?

“When people are confused, they say no.”

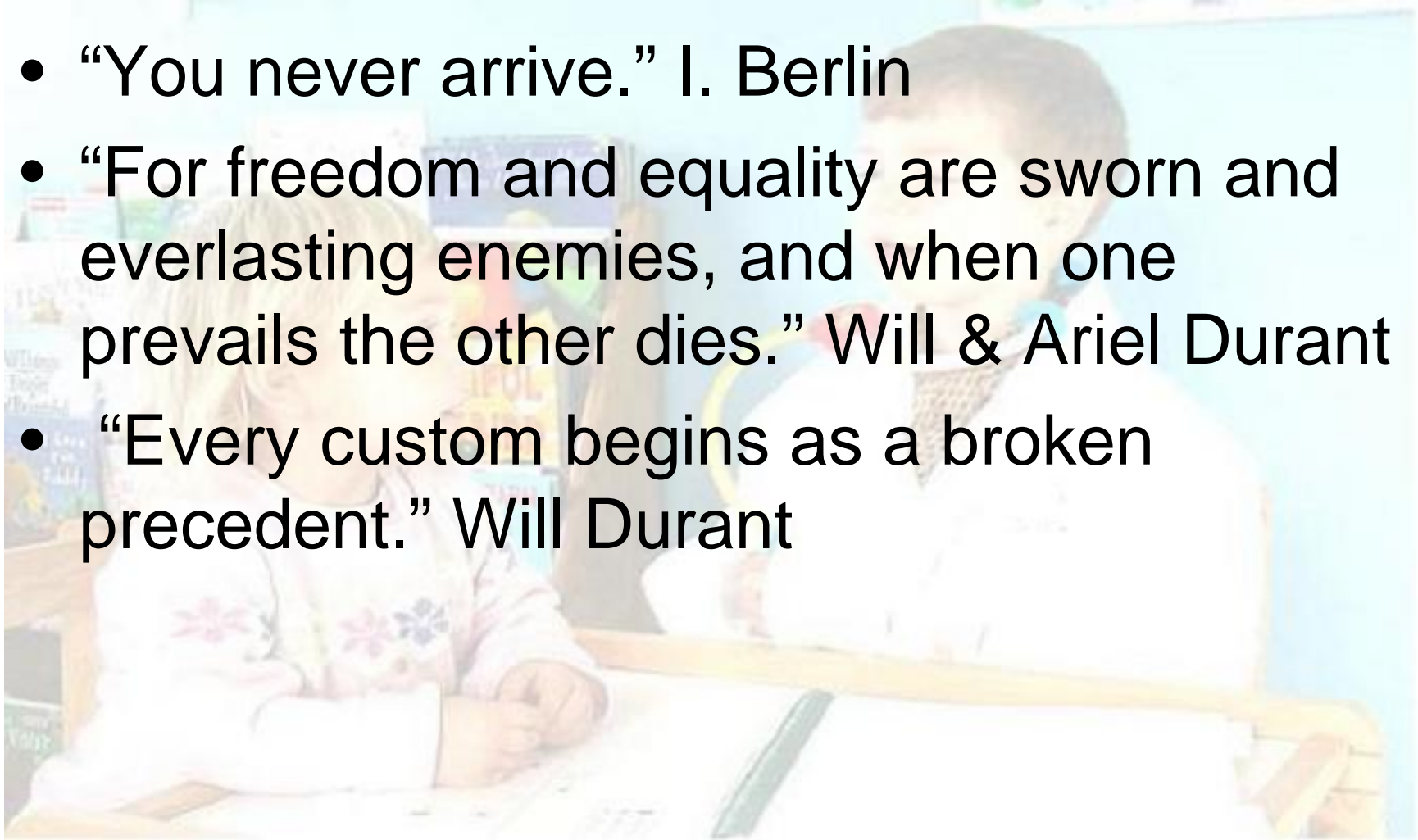
Family history “tools” over time

- Adam and Eve begat Cain and Abel
- Royal pedigrees important for succession and land distribution
- Percent “race”, ancestry profiles, degrees of relationship important for freedom, legal rights, inheritance, place in baby book, wedding invitations, transplantation, advance directives



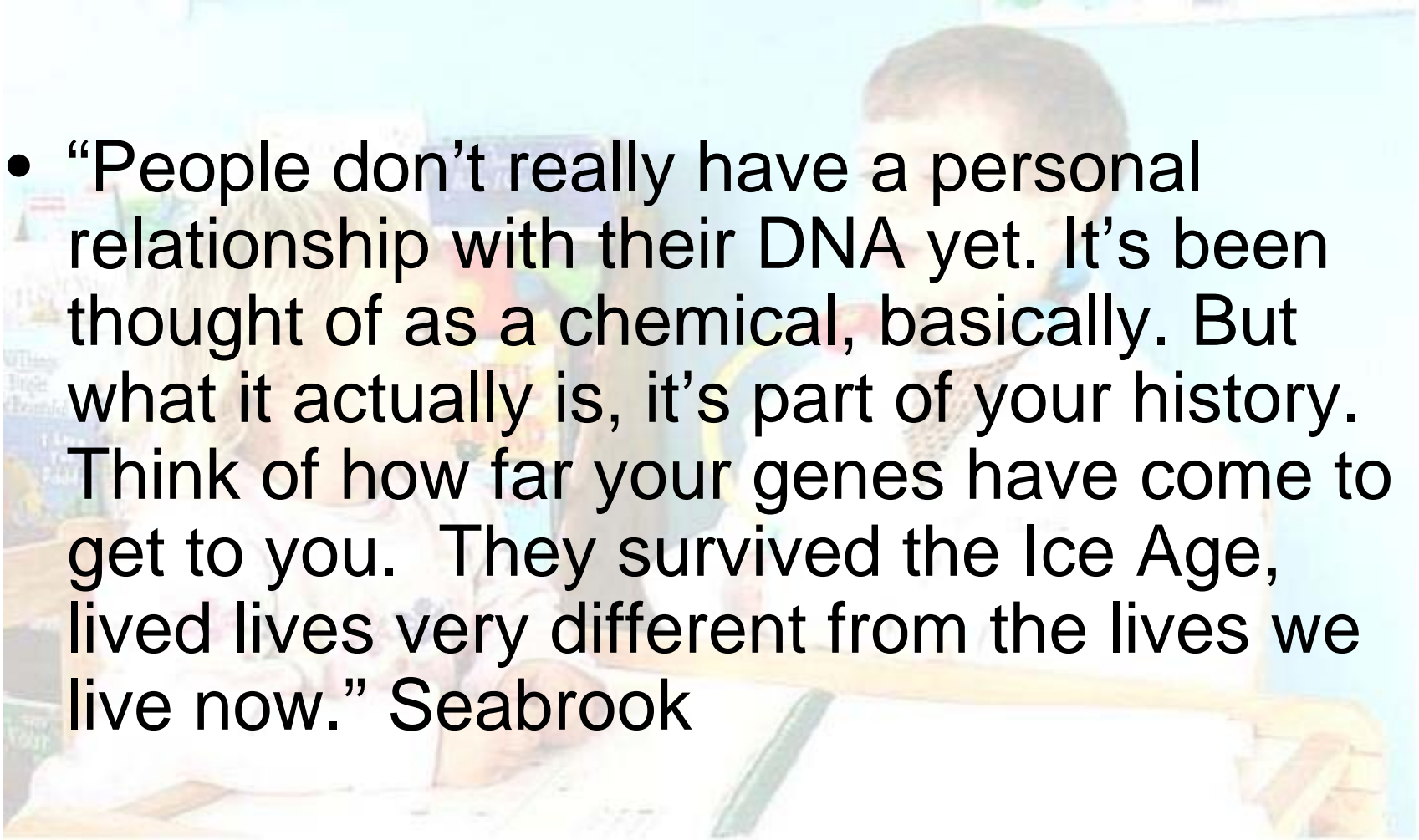
Family history as a societal priority - entertainment??

- “You never arrive.” I. Berlin
- “For freedom and equality are sworn and everlasting enemies, and when one prevails the other dies.” Will & Ariel Durant
- “Every custom begins as a broken precedent.” Will Durant



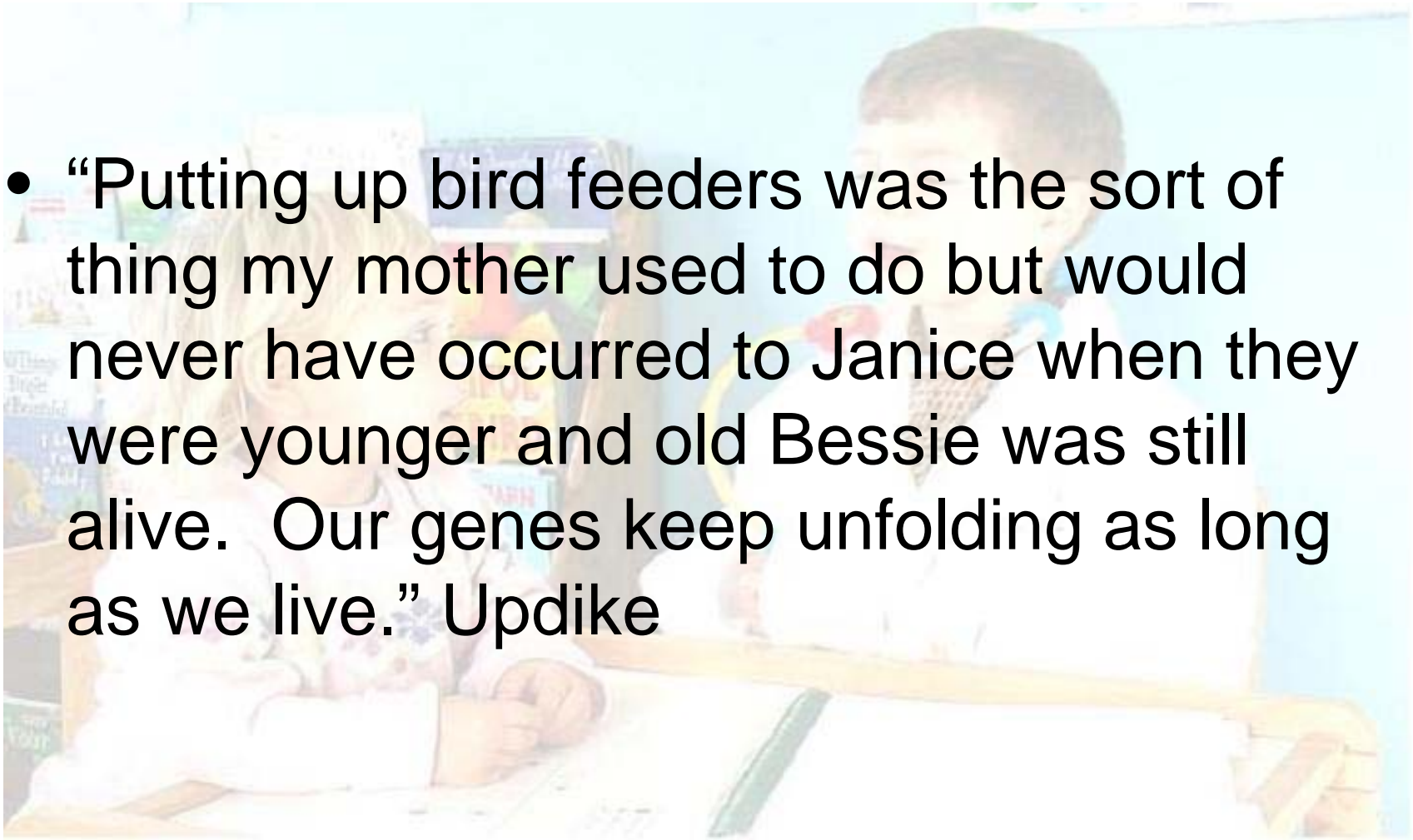
Perspectives

- “People don’t really have a personal relationship with their DNA yet. It’s been thought of as a chemical, basically. But what it actually is, it’s part of your history. Think of how far your genes have come to get to you. They survived the Ice Age, lived lives very different from the lives we live now.” Seabrook



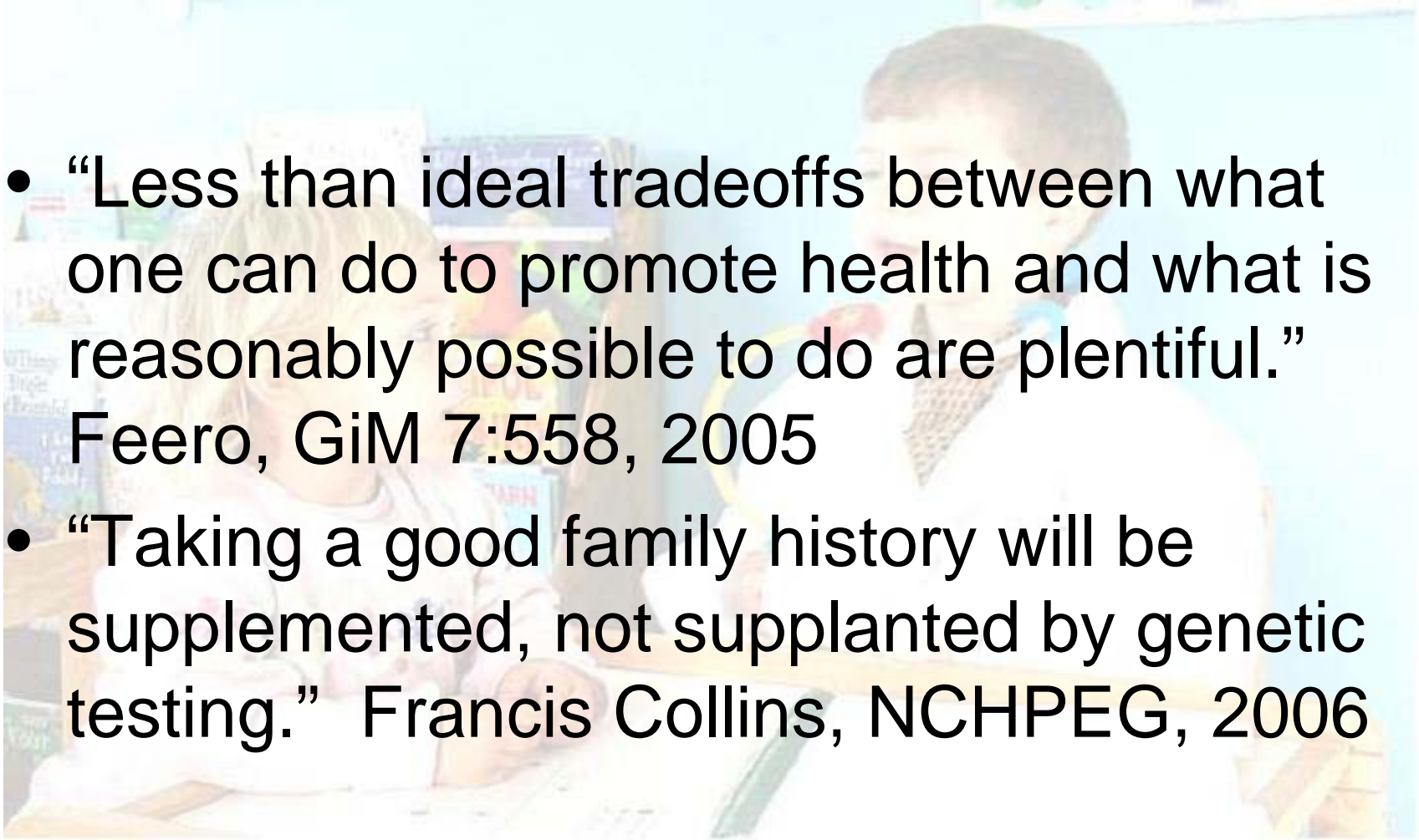
Perspectives

- “Putting up bird feeders was the sort of thing my mother used to do but would never have occurred to Janice when they were younger and old Bessie was still alive. Our genes keep unfolding as long as we live.” Updike



Perspectives

- “Less than ideal tradeoffs between what one can do to promote health and what is reasonably possible to do are plentiful.”
Feero, GiM 7:558, 2005
- “Taking a good family history will be supplemented, not supplanted by genetic testing.” Francis Collins, NCHPEG, 2006

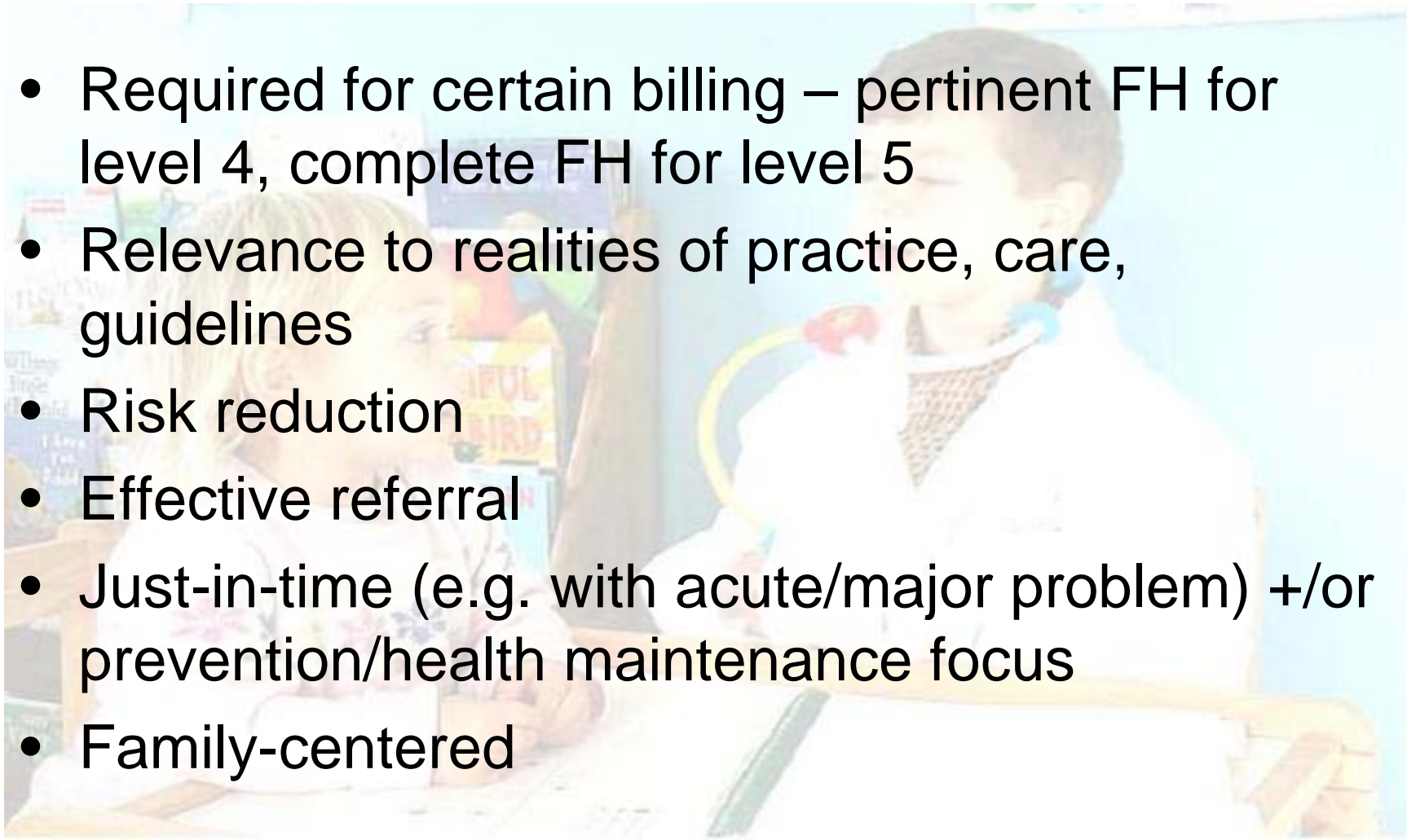


Conceptualizing

- **Child in family context** – genealogy, family stories/keepsakes/caring, medical records vs. unknown parentage, unreliable/absent records, parental rights
- **Child as autonomous** – newborn screen, genetic tests, medical records, transition to adulthood
- **Child as individual in records systems** – baby books, birth certificates, registries, medical home, immunization tracking, acute care, hospitalization, EMR
- **Child as consumer** – marketable FH products, drug marketing, risk/litigation avoidance, quality assurance, healthy adult
- **Child as “special”, “with disposition”, “marked”, high risk**

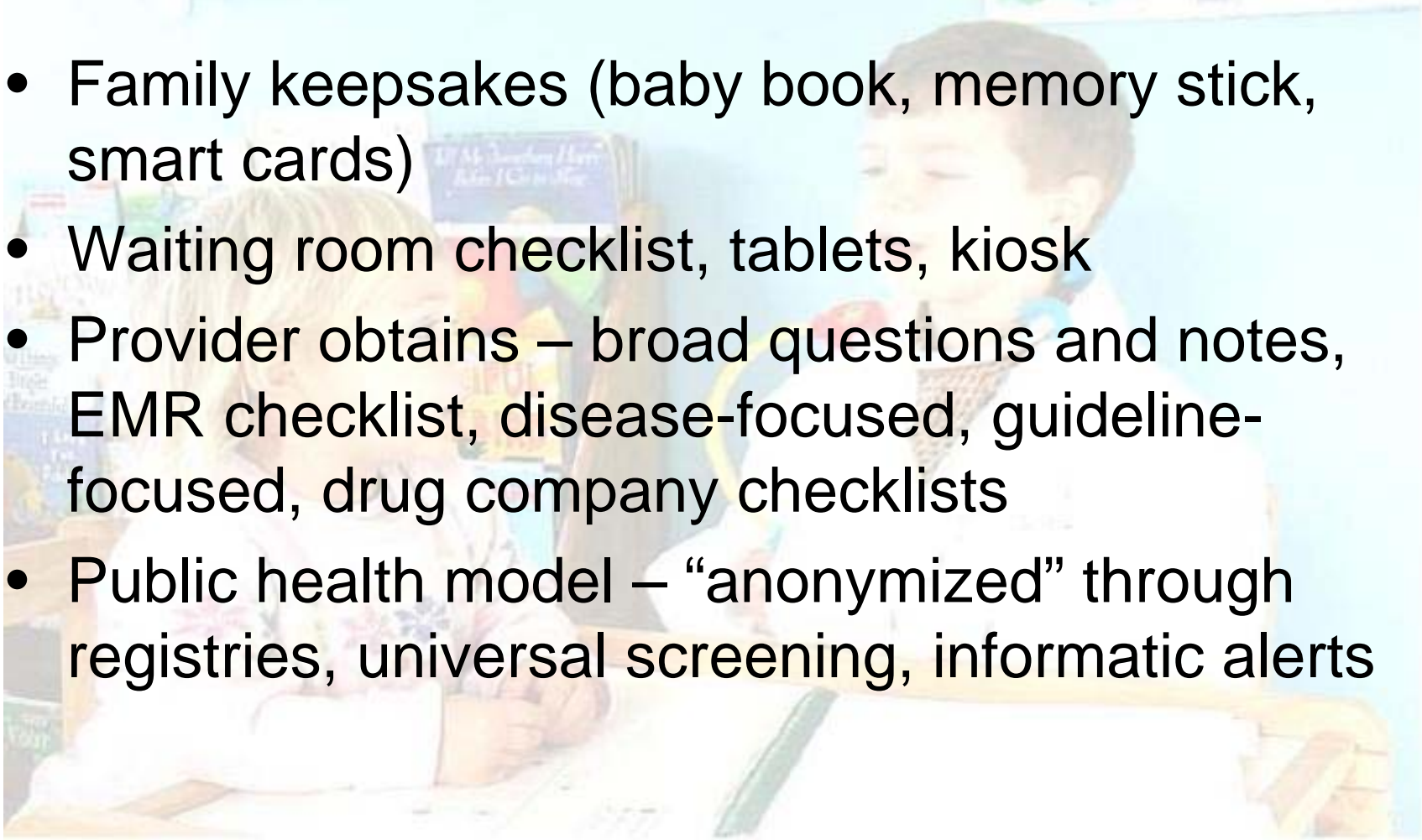
FH Needs of Pediatricians

- Required for certain billing – pertinent FH for level 4, complete FH for level 5
- Relevance to realities of practice, care, guidelines
- Risk reduction
- Effective referral
- Just-in-time (e.g. with acute/major problem) +/- prevention/health maintenance focus
- Family-centered



Varied approaches to FH

- Family keepsakes (baby book, memory stick, smart cards)
- Waiting room checklist, tablets, kiosk
- Provider obtains – broad questions and notes, EMR checklist, disease-focused, guideline-focused, drug company checklists
- Public health model – “anonymized” through registries, universal screening, informatic alerts



The lay of the land

- **AMA*** (www.ama-asn.org Pediatric Clinical Genetics Questionnaire) pocket card
- **Surgeon General*** (www.hhs.gov/familyhistory/)
- **Genzyme** (www.fabrycommunity.com)
- **MOD*** (www.marchofdimes.com/files/FamilyHealthHistory.pdf)
- **Mayo Clinic** (www.mayoclinic.com/health/medical-history)
- **NSGC** (www.nsgc.org/consumer/familytree)
- **Bright Futures** (www.brightfutures.org/mentalhealth/pdf/professionals/ped_intake_form.pdf)

Several drug companies with minimal attention to pediatric issues, CDC website

- ***Bring to MD/genetic counselor**

AMA Pediatric Checklist

- **Pedigree**
(Family History Tree)
- [Click to see a sample pedigree in PowerPoint](#)



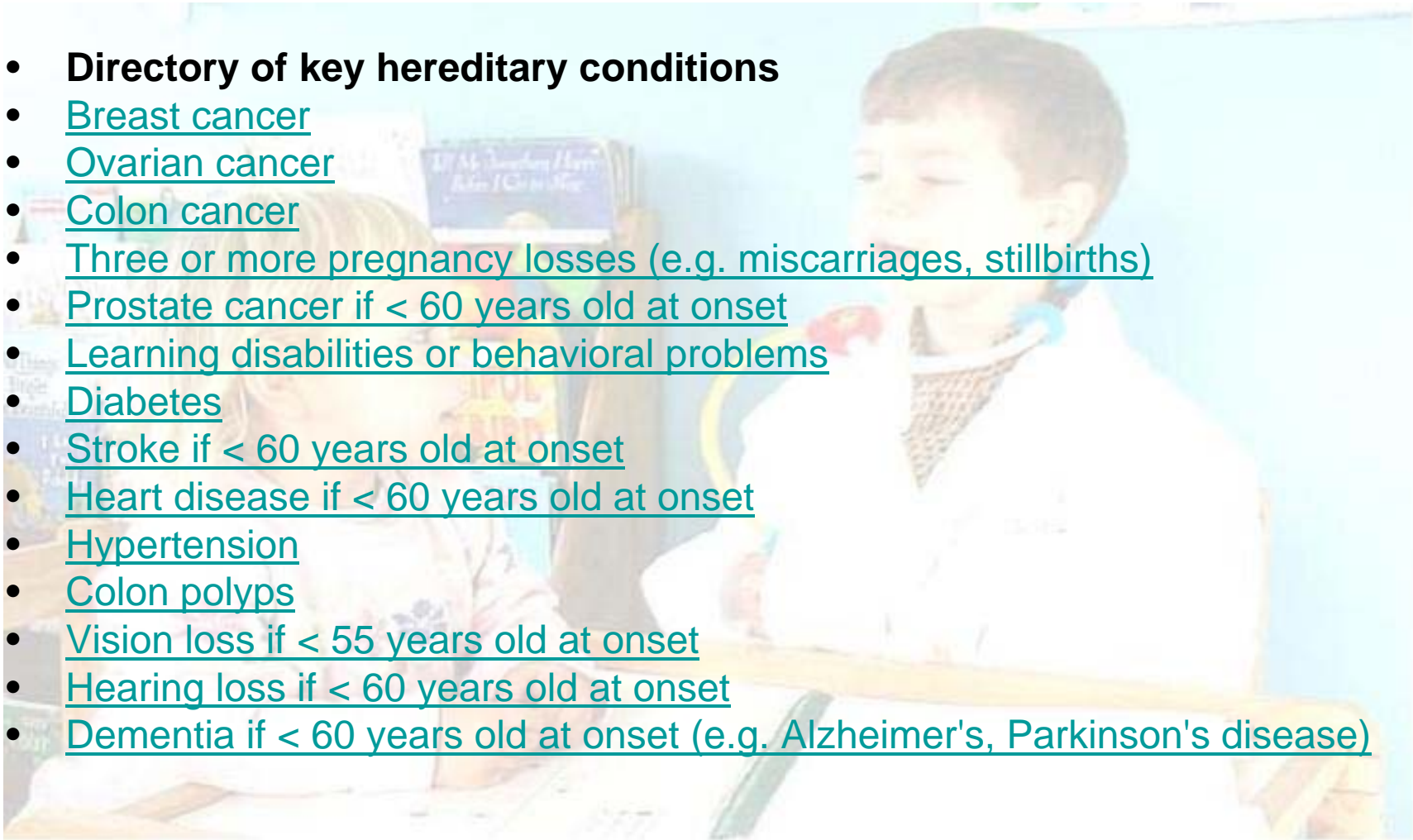
Surgeon General List

- Heart disease
- Stroke
- Diabetes
- Breast Cancer
- Colon Cancer
- Ovarian Cancer



Norwich Union Health Tree

- **Directory of key hereditary conditions**
- [Breast cancer](#)
- [Ovarian cancer](#)
- [Colon cancer](#)
- [Three or more pregnancy losses \(e.g. miscarriages, stillbirths\)](#)
- [Prostate cancer if < 60 years old at onset](#)
- [Learning disabilities or behavioral problems](#)
- [Diabetes](#)
- [Stroke if < 60 years old at onset](#)
- [Heart disease if < 60 years old at onset](#)
- [Hypertension](#)
- [Colon polyps](#)
- [Vision loss if < 55 years old at onset](#)
- [Hearing loss if < 60 years old at onset](#)
- [Dementia if < 60 years old at onset \(e.g. Alzheimer's, Parkinson's disease\)](#)

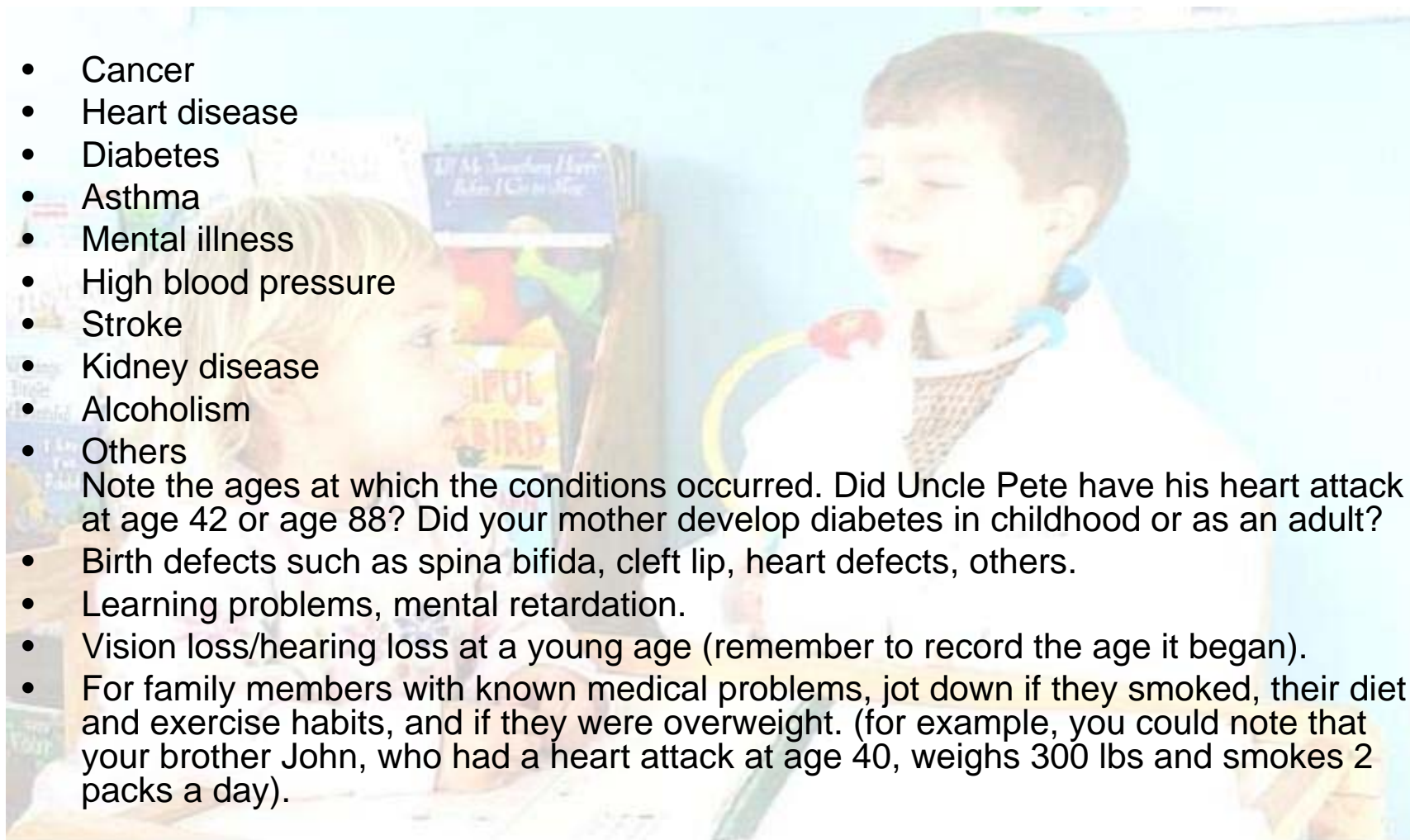


NSGC list

- Cancer
- Heart disease
- Diabetes
- Asthma
- Mental illness
- High blood pressure
- Stroke
- Kidney disease
- Alcoholism
- Others

Note the ages at which the conditions occurred. Did Uncle Pete have his heart attack at age 42 or age 88? Did your mother develop diabetes in childhood or as an adult?

- Birth defects such as spina bifida, cleft lip, heart defects, others.
- Learning problems, mental retardation.
- Vision loss/hearing loss at a young age (remember to record the age it began).
- For family members with known medical problems, jot down if they smoked, their diet and exercise habits, and if they were overweight. (for example, you could note that your brother John, who had a heart attack at age 40, weighs 300 lbs and smokes 2 packs a day).



Bright Futures

- 25 separate pages with some reference or potential link to FH +/- or genetic differences
- Suggests 3 generation health and social history, including congenital disabilities and genetic disorders
- Family-centered prenatal visit – “Are you concerned that your baby will inherit any diseases or characteristics that run in the family?” But no pedigree or “If so, have you had genetic counseling?”
- FH form under discussion. Currently a few FH questions being considered for new forms.

Family History

Have any family members had the following:

- | | | | | |
|---|------------------------------|-----------------------------|-----------|----------------|
| Deafness | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Nasal allergies | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Asthma | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Tuberculosis | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Heart disease (before 50 years old) | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| High blood pressure (before 50 years old) | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| High cholesterol | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Anemia | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Bleeding disorder | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Liver disease | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Kidney disease | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Diabetes (before 50 years old) | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Bed-wetting (after 10 years old) | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Epilepsy or convulsions | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Alcohol abuse | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Drug abuse | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Mental illness | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Mental retardation | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |
| Immune problems, HIV, or AIDS | <input type="checkbox"/> Yes | <input type="checkbox"/> No | Who _____ | Comments _____ |

Additional family history _____

VCU Dept. of Pediatrics Initial Visit

FAMILY MEDICAL HISTORY:
Please circle the problem if it runs in the family and note who has the problem (by number) in the space below.

1. Alcohol/drug problems	6. Cancer	11. High cholesterol	16. Mental retardation	21. Die as an infant?
2. Asthma	7. Deafness	12. HIV/AIDS	17. Seizures	22. Other diseases?
3. Birth defects	8. Diabetes	13. Hyperactivity	18. Skin problems	
4. Blindness	9. Heart disease	14. Learning disabilities	19. Tuberculosis	
5. Blood diseases (sickle cell)	10. High blood pressure	15. Mental illness	20. Die younger than 50 years old?	

Notes:

- What should be on the list? What to do with information?

Name: _____		Room: _____	
DOB: _____ MRN: _____		MCV Hospitals and Physicians VCU Health System Richmond, Virginia 23298	
Date: ____/____/____		PEDIATRIC HEALTH MAINTENANCE NEWBORN VISIT - Age of Child: _____	
CONCERNS:		Source: _____	
PAIN ASSESSMENT: Pain? Y / N Tool: Number / Faces / Flacc Score: _____ Where? _____ LPN/RN Sign: _____			
BIRTH HISTORY Gestation: _____ wks Birth weight: _____ lbs _____ oz Mom's age: _____ y/o Mom's G: _____ P: _____ A: _____ PNC: _____ GBS HepB RPR HIV GC CN		PERINATAL HISTORY Length of hospital stay: _____ D/C weight: _____ Complications: <input type="checkbox"/> None <input type="checkbox"/> Other: _____	
FAMILY HISTORY <input type="checkbox"/> See New Patient Questionnaire		SOCIAL HISTORY • Who lives at home? • Social stressors/supports?	
Medications: <input type="checkbox"/> None <input type="checkbox"/> Other: _____		Diet: _____	
Allergies: <input type="checkbox"/> NKDA <input type="checkbox"/> Other: _____		Elimination: _____ Sleep (hrs/position): _____	
REVIEW OF SYSTEMS: General Sx <input type="checkbox"/> N <input type="checkbox"/> Abn: Eyes <input type="checkbox"/> N <input type="checkbox"/> Abn: ENT <input type="checkbox"/> N <input type="checkbox"/> Abn: CV <input type="checkbox"/> N <input type="checkbox"/> Abn: Resp <input type="checkbox"/> N <input type="checkbox"/> Abn: GI <input type="checkbox"/> N <input type="checkbox"/> Abn: GU <input type="checkbox"/> N <input type="checkbox"/> Abn: Musc/skel <input type="checkbox"/> N <input type="checkbox"/> Abn: Skin/breast <input type="checkbox"/> N <input type="checkbox"/> Abn: Neuro <input type="checkbox"/> N <input type="checkbox"/> Abn: Psych <input type="checkbox"/> N <input type="checkbox"/> Abn: Endocrine <input type="checkbox"/> N <input type="checkbox"/> Abn: Hem/Lymph <input type="checkbox"/> N <input type="checkbox"/> Abn: Aller/Imm <input type="checkbox"/> N <input type="checkbox"/> Abn: <input type="checkbox"/> Complete ROS performed, pertinent findings noted		EXAMINATION: Wt: (_____ lbs) Ht: (_____ in) HC: (_____ cm) Wt: (_____ kg) Ht: (_____ cm) HC: (_____ cm) Temp: (_____); Resp: (_____); Pulse: (_____); Blood Pressure (_____ / _____) General: <input type="checkbox"/> N <input type="checkbox"/> Abn: Skin: <input type="checkbox"/> N <input type="checkbox"/> Abn: Head: <input type="checkbox"/> N <input type="checkbox"/> Abn: Eyes: <input type="checkbox"/> N <input type="checkbox"/> Abn: Ears: <input type="checkbox"/> N <input type="checkbox"/> Abn: Nose/Throat/Neck: <input type="checkbox"/> N <input type="checkbox"/> Abn: Chest/Lungs: <input type="checkbox"/> N <input type="checkbox"/> Abn: Heart/Pulses: <input type="checkbox"/> N <input type="checkbox"/> Abn: Abdomen: <input type="checkbox"/> N <input type="checkbox"/> Abn: GU: <input type="checkbox"/> N <input type="checkbox"/> Abn: Extremities: <input type="checkbox"/> N <input type="checkbox"/> Abn: Neurologic: <input type="checkbox"/> N <input type="checkbox"/> Abn: Development: <input type="checkbox"/> N <input type="checkbox"/> Abn:	
ANTICIPATORY GUIDANCE: Barriers: N / L / CR / P / C / S (N=None L=Language CR=Cultural/Religious P=Physical Impairment C=Cognitive Limitation S=Psychosocial) <input type="checkbox"/> Car seat <input type="checkbox"/> Smoking/Smoke alarms <input type="checkbox"/> Fevers <input type="checkbox"/> No water <input type="checkbox"/> Maternal fatigue/return to work <input type="checkbox"/> Shaken Baby <input type="checkbox"/> Hydration <input type="checkbox"/> Sleep position <input type="checkbox"/> Respiratory Status <input type="checkbox"/> Water temp <input type="checkbox"/> Vitamin D Supplementation <input type="checkbox"/> Normal development/immunizations <input type="checkbox"/> Policy on calls, ER, and when to call reviewed? (yes/no)			
DIAGNOSIS/PLAN:		NEWBORN SCREEN: <input type="checkbox"/> N <input type="checkbox"/> Other: _____	
ATTENDING DOCUMENTATION:		LABS: (None indicated) Other: _____	
<input type="checkbox"/> I was present with the Resident during the entire interview and examination of the patient. I repeated the key portions of the exam in the presence of the Resident. I confirmed/revised the Resident's History, Exam, Assessment and Plan as noted above. See Resident's notes for details. <input type="checkbox"/> I was NOT present with the Resident during the entire interview and examination of the patient. I personally interviewed the patient and repeated the examination. I confirmed/revised the Resident's History, Exam, Assessment and Plan as noted above. See Resident's notes for details.		IMMUNIZATIONS: (none indicated) <input type="checkbox"/> HepB # 1 (if not done in hospital): Other: _____	
RESIDENT SIGNATURE: _____	DATE: _____	ATTENDING SIGNATURE: _____	DATE: _____

CHRONIC DIAGNOSIS/HISTORY See Progress Notes**Pt. Family**

- | | | |
|--------------------------|--------------------------|--------------------------|
| <input type="checkbox"/> | <input type="checkbox"/> | Arthritis |
| <input type="checkbox"/> | <input type="checkbox"/> | Asthma |
| <input type="checkbox"/> | <input type="checkbox"/> | Cancer |
| <input type="checkbox"/> | <input type="checkbox"/> | Congestive Heart Failure |
| <input type="checkbox"/> | <input type="checkbox"/> | Depression |
| <input type="checkbox"/> | <input type="checkbox"/> | Other _____ |

Pt. Family

- | | | |
|--------------------------|--------------------------|---------------|
| <input type="checkbox"/> | <input type="checkbox"/> | Diabetes |
| <input type="checkbox"/> | <input type="checkbox"/> | Stroke |
| <input type="checkbox"/> | <input type="checkbox"/> | Hypertension |
| <input type="checkbox"/> | <input type="checkbox"/> | Seizures |
| <input type="checkbox"/> | <input type="checkbox"/> | Renal Failure |

Family History:

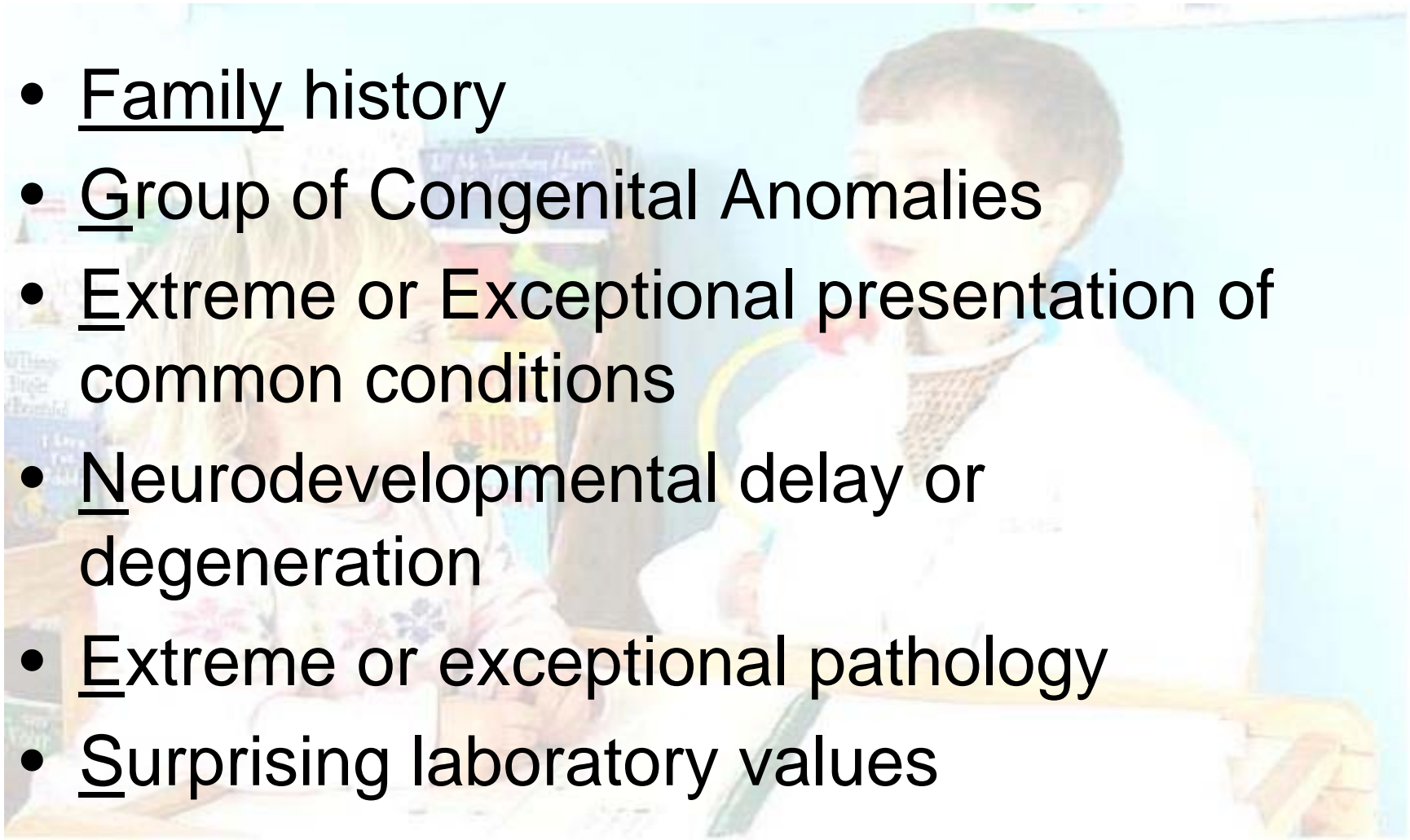
- | | | | |
|-----------|---------------------------------|-----------------------------------|----------------------------------|
| Mother: | <input type="checkbox"/> Living | <input type="checkbox"/> Deceased | <input type="checkbox"/> Unknown |
| Father: | <input type="checkbox"/> Living | <input type="checkbox"/> Deceased | <input type="checkbox"/> Unknown |
| Siblings: | <input type="checkbox"/> Living | <input type="checkbox"/> Deceased | <input type="checkbox"/> Unknown |

Past Surgical/Hospitalization History: _____

OB - G _____ P _____ A _____ Date of last menses: _____

GPC Initiative

- Family history
- Group of Congenital Anomalies
- Extrême or Exceptional presentation of common conditions
- Neurodevelopmental delay or degeneration
- Extrême or exceptional pathology
- Surprising laboratory values

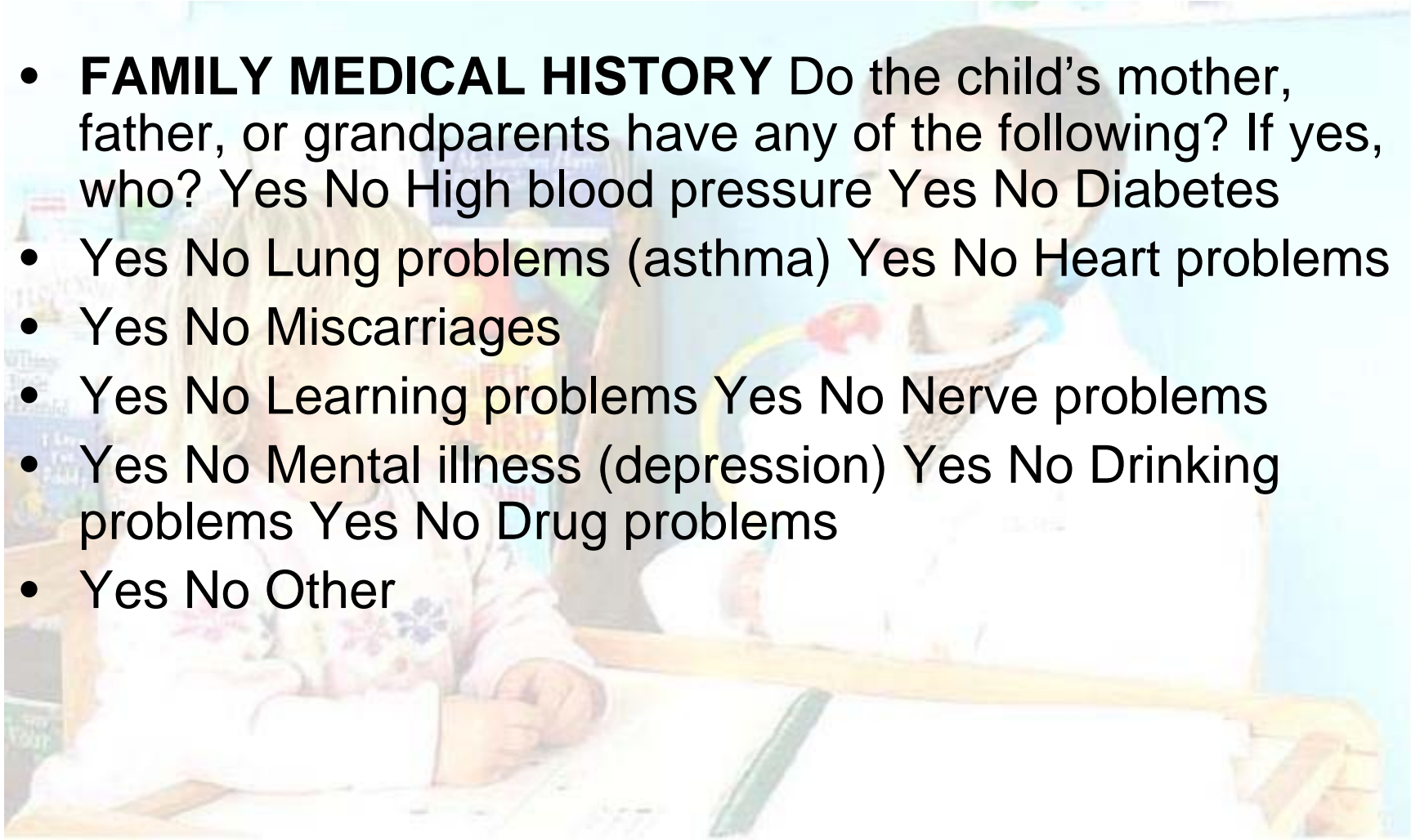


Some observations

- Forms (e.g. templates) trump function.
7 of 11 templates at our hospital with FH space/checklist; 23/30 charts with some info.
- Time (e.g. 6 peds pts/hr) trumps completeness.
- Outliers (e.g. young deaths, discrimination stories) trump routine maintenance.
- Technology (e.g. alerts, structured lists) can help.
- Models (e.g. E&M comprehensive billing, health belief model, child – “adult to be”) may help.

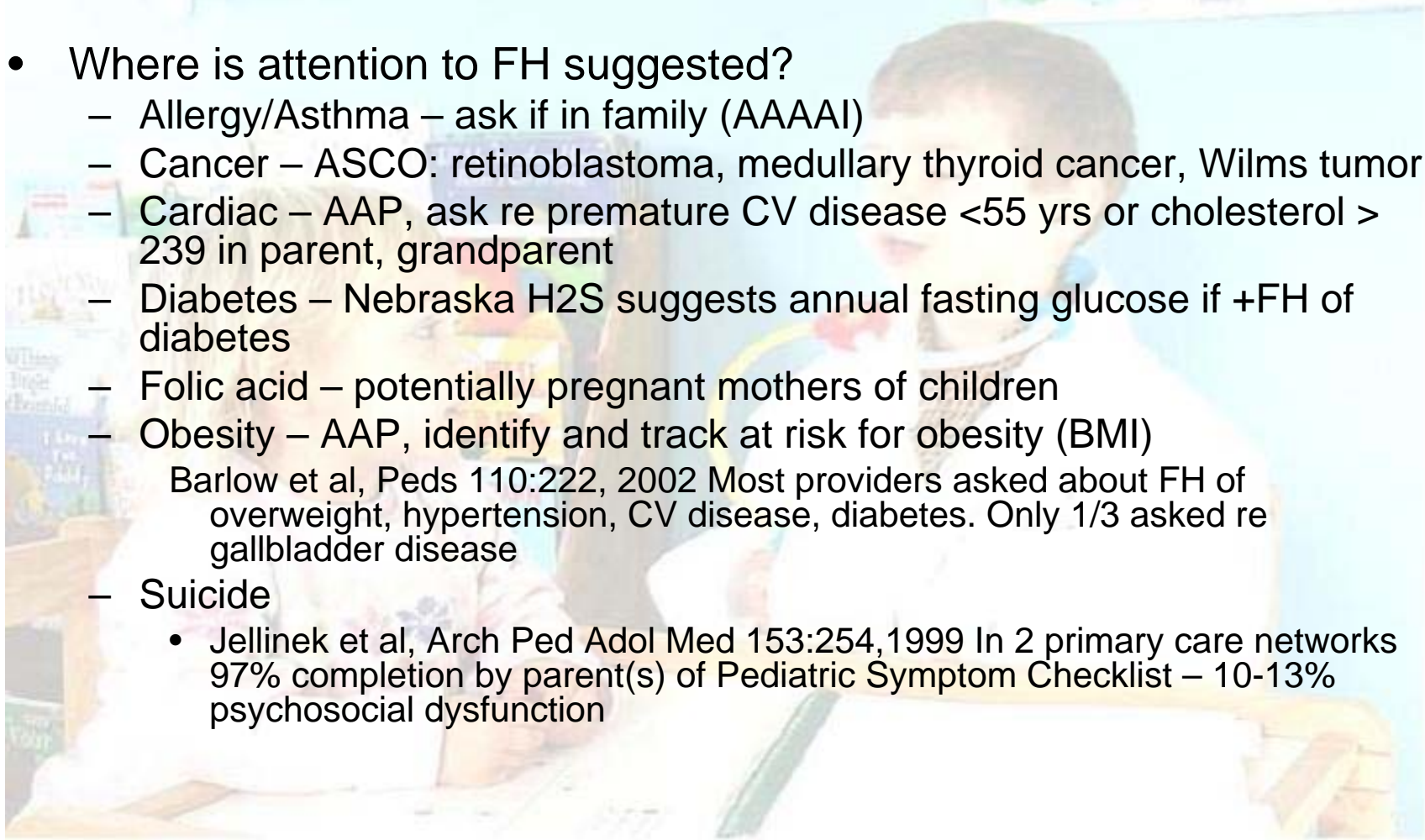
Kemper's Pediatric Mental Health Checklist

- **FAMILY MEDICAL HISTORY** Do the child's mother, father, or grandparents have any of the following? If yes, who? Yes No High blood pressure Yes No Diabetes
- Yes No Lung problems (asthma) Yes No Heart problems
- Yes No Miscarriages
- Yes No Learning problems Yes No Nerve problems
- Yes No Mental illness (depression) Yes No Drinking problems Yes No Drug problems
- Yes No Other



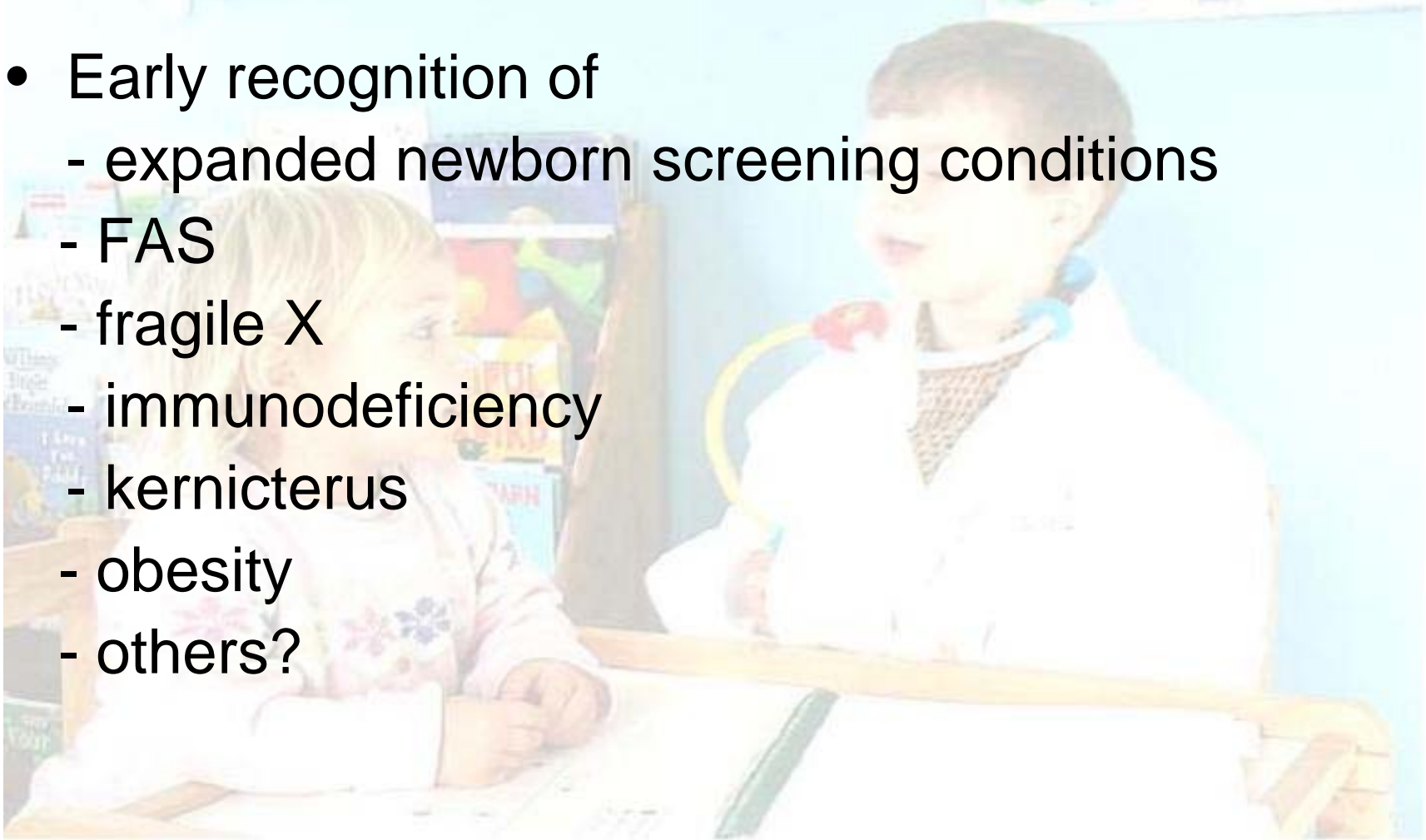
Pediatric Guidelines

- Where is attention to FH suggested?
 - Allergy/Asthma – ask if in family (AAAAI)
 - Cancer – ASCO: retinoblastoma, medullary thyroid cancer, Wilms tumor
 - Cardiac – AAP, ask re premature CV disease <55 yrs or cholesterol > 239 in parent, grandparent
 - Diabetes – Nebraska H2S suggests annual fasting glucose if +FH of diabetes
 - Folic acid – potentially pregnant mothers of children
 - Obesity – AAP, identify and track at risk for obesity (BMI)
 - Barlow et al, Peds 110:222, 2002 Most providers asked about FH of overweight, hypertension, CV disease, diabetes. Only 1/3 asked re gallbladder disease
 - Suicide
 - Jellinek et al, Arch Ped Adol Med 153:254,1999 In 2 primary care networks 97% completion by parent(s) of Pediatric Symptom Checklist – 10-13% psychosocial dysfunction



National pediatric concerns

- Early recognition of
 - expanded newborn screening conditions
 - FAS
 - fragile X
 - immunodeficiency
 - kernicterus
 - obesity
 - others?

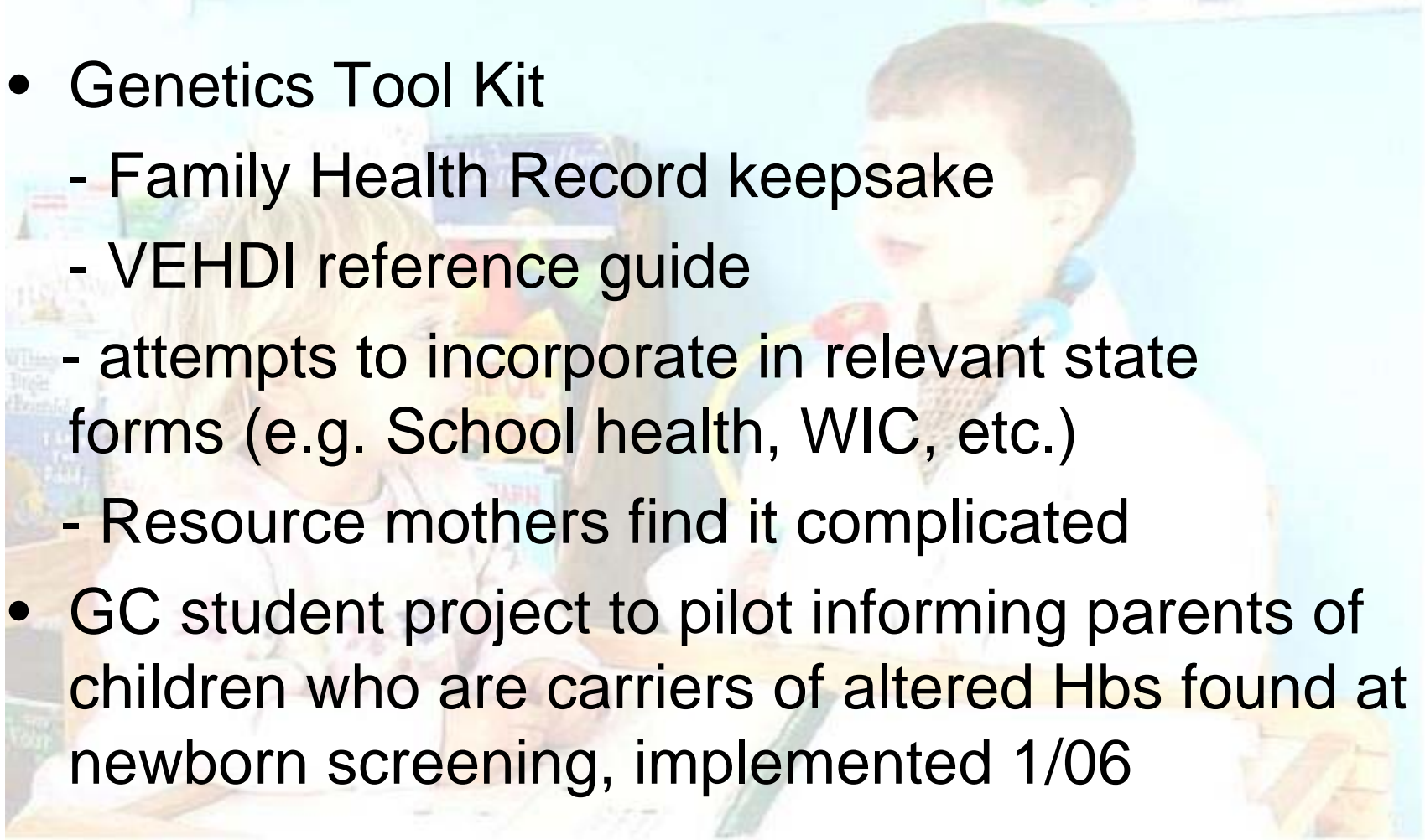


Put it on the list

- Gritz et al, Ped Derm 20:16,2003 -76% recommend sunscreen, ~22% take FH
- Hampel et al, NEJM 352:1851,2005 – offer mutation testing with Lynch syndrome in relative with CRC
- Multiple and varied suggestions and recommendations given specific signs and symptoms (no specific USPSTF pediatric recommendations except not enough evidence for newborn hearing screening)

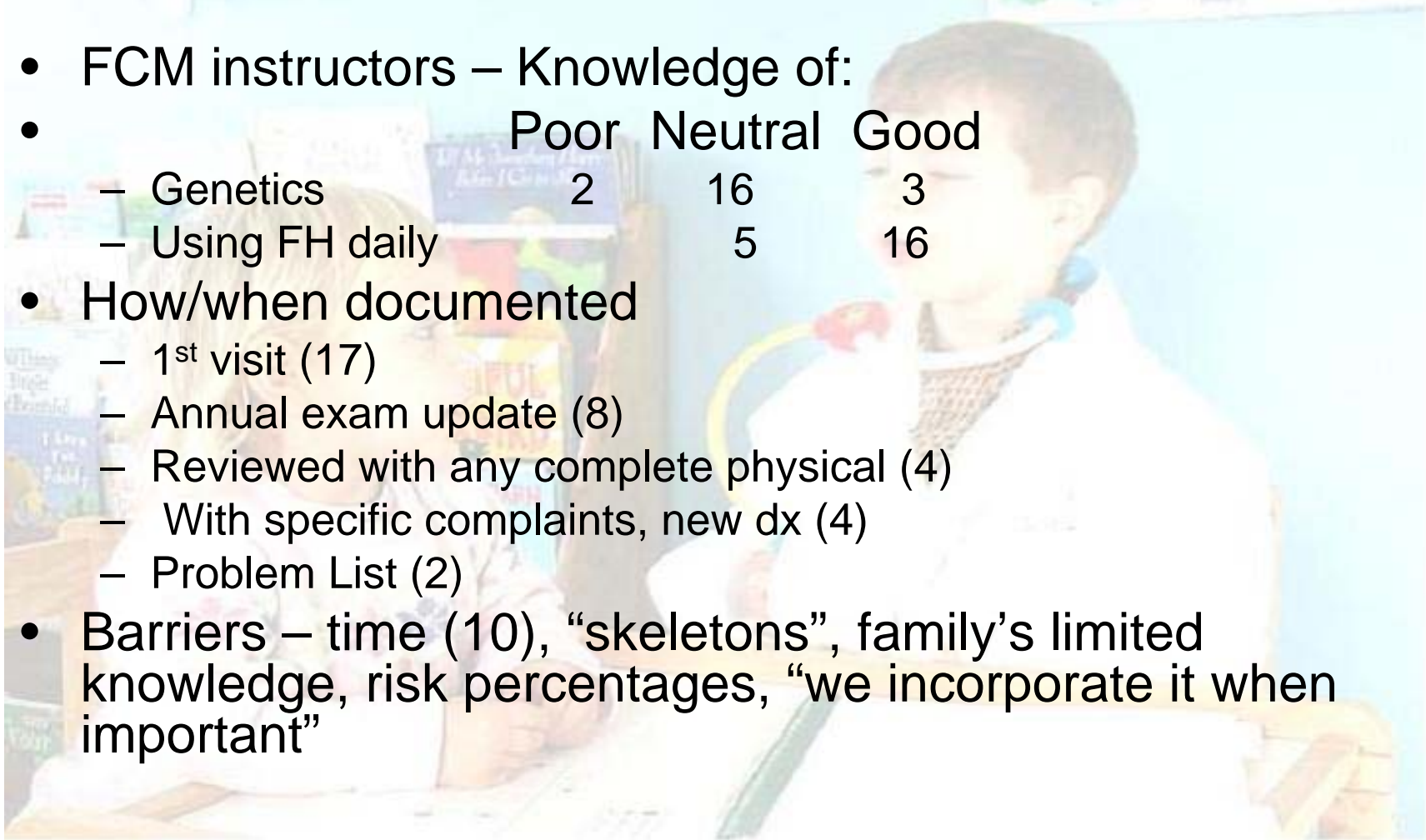
Virginia experience – state level

- Genetics Tool Kit
 - Family Health Record keepsake
 - VEHDI reference guide
 - attempts to incorporate in relevant state forms (e.g. School health, WIC, etc.)
 - Resource mothers find it complicated
- GC student project to pilot informing parents of children who are carriers of altered Hbs found at newborn screening, implemented 1/06



Virginia experience - instruction

- FCM instructors – Knowledge of:
- | | Poor | Neutral | Good |
|------------------|------|---------|------|
| – Genetics | 2 | 16 | 3 |
| – Using FH daily | | 5 | 16 |
- How/when documented
 - 1st visit (17)
 - Annual exam update (8)
 - Reviewed with any complete physical (4)
 - With specific complaints, new dx (4)
 - Problem List (2)
- Barriers – time (10), “skeletons”, family’s limited knowledge, risk percentages, “we incorporate it when important”



Virginia experience - VCUHS

- 1999 – family medical hx in 91/165 (55%) of charts
- Cerner discussions, pharmacogenomics initiative, asthma med and coumadin metabolism testing
- Bariatric surgery discussions
- Cancer – patient form not yet integrated in hospital record
- Cardiovascular risk
- BLANK space in new template
- Pediatric waiting room tool to be piloted

SIDE questions

- Mother's side

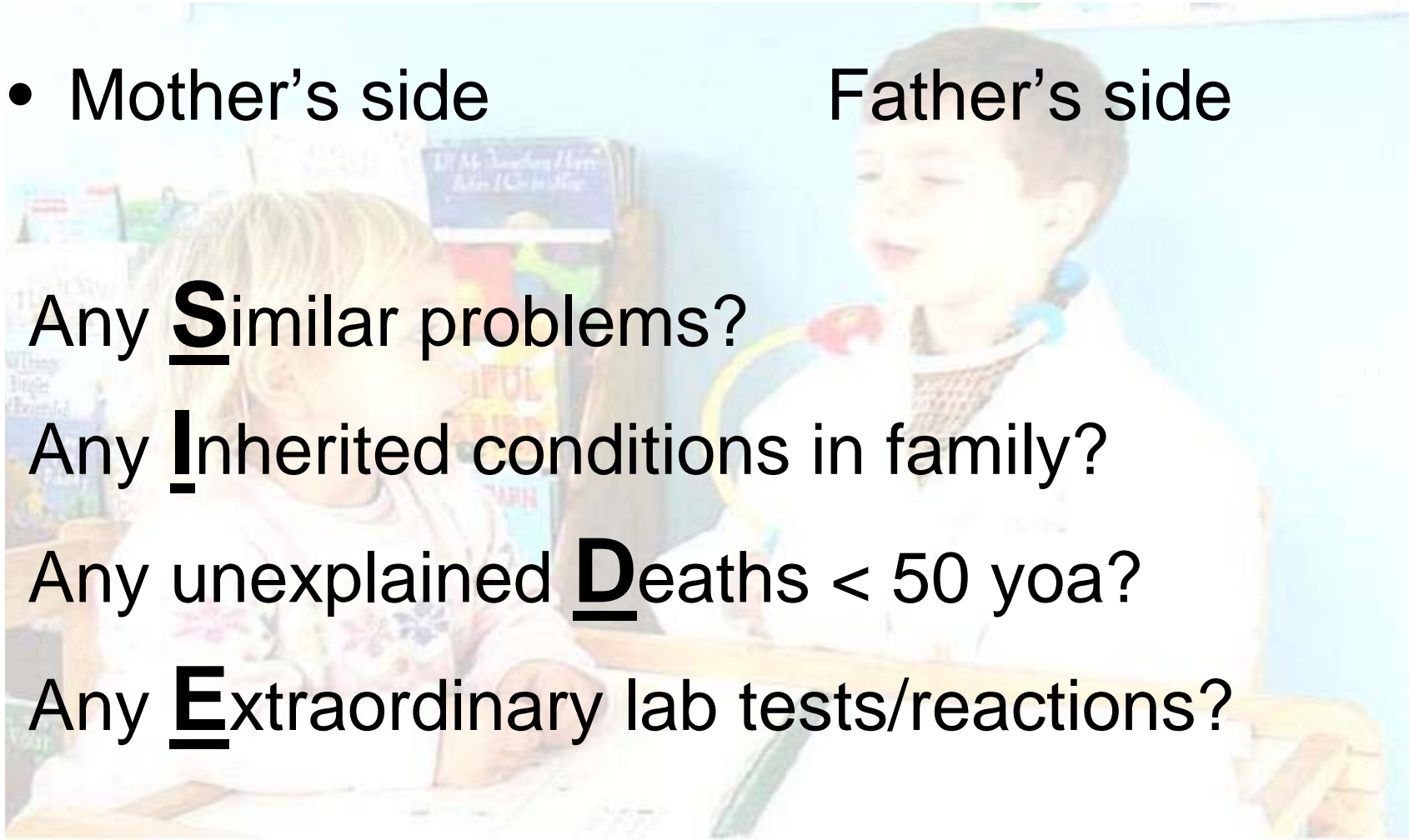
Father's side

Any **S**imilar problems?

Any **I**nherited conditions in family?

Any unexplained **D**eaths < 50 yoa?

Any **E**xtraordinary lab tests/reactions?



Family history

- Lost in translation?



Goldilocks (“just right”) solutions

- Encourage sharing of family health history within families and documentation
- Pediatrician to have FH checklist for first visit/birth and annual update
- SIDE questions for hospitalization and major unexplained illness
- ?Refer any child to genetics with 2 or more relatives with similar problems, child or <50 yoa death, abnormal genetic test