What is Genetic Counseling?

- Genetic counseling is the practice of helping individuals and families understand the medical, psychological, social and reproductive implications of genetic and congenital conditions.
- Elements of the practice include: Assessment of the chance for recurrence or occurrence of a condition, Education about inheritance, testing options, medical management, prevention, social support and research, and Counseling to help clients adapt to the choices and to the psychological, familial and social issues that stem from the risk or condition in the family.
What is a Genetic Counselor?

- Genetic counselors work as members of a health care team providing genetic services.
- Most have a Master’s degree in Genetics or Genetic Counseling with an undergraduate degree in a biological science.
- Genetic counselors are board-certified and may be licensed in the near future.
Genetic Centers in NC

- Clinical Centers: Charlotte, Asheville, Winston-Salem, Durham, Chapel Hill, Greensboro, Greenville, Wilmington
- Private/Commercial: Chapel Hill, RTP, reference labs (LabCorp, Athena, Myriad, ...) , others
- Public Health GCs (work for state of NC)
Roles for Genetic Counselors

- Prenatal
- Pediatric
- Adult
- Cancer
- Neuromuscular
- Research
- Commercial
- Private Practice
- Create your own specialty – the field is wide open!!
“There you go, Mrs. Eagen — you can clearly see both twins on the monitor.”
Prenatal Genetic Counseling

- Advanced Maternal Age (age > 35)
- Screening tests indicating an increased risk
- Family history questions
- Teratogenic exposure
- Suspicious ultrasound findings
Prenatal Diagnostic Tests Offered

- Level II ultrasound
- Chorionic Villus Sampling (CVS)
- Amniocentesis
- Percutaneous Umbilical Blood Sampling (PUBS)
Risks of Procedures

- Amniocentesis: risk of complications, including miscarriage, increased by 0.5%. Risk of 1/200 quoted but may be more like 1/500 in experienced hands.
- CVS: risk of complications, including miscarriage, increased up to 1%.
Risk Perception

Is a 1% risk considered a “high” risk or a “low” risk??
Non-directive counseling (incredibly simplified)

- Information and options given and explored but couples never told they must have testing or what to do with the results.
- Never answer the question “what would you do if you were me?”
- Fortunately, 98% of tests show normal results
Baseline Pregnancy Risks

- 3-5% background risk to have a child born with some type of birth defect or mental retardation (REGARDLESS of prenatal care, family history, etc.)
- Each one of us carry anywhere from 5 to 10 recessive genes - we are ALL carriers for something!
- NO TEST CAN GUARANTEE A PERFECT BABY!!!
Dirk brings his family tree to class
Indications for Genetic Evaluation

- Birth defects
- Developmental delays or mental retardation
- Distinctive features
- Known family history of genetic condition
- Unusual genetic or biochemical testing results
The Genetic Evaluation

- Complete medical & developmental hx
- Physical exam
- Careful family hx
- Laboratory/imaging studies
- Genetic counseling
- Management & referrals
Giving Unexpected News

- Important to proceed tactfully, truthfully, and empathetically
- Emphasize what the child will be able to do, not what they will not!
- Remind parents to treat this baby like their other children and to have expectations for this child
- This child will be more LIKE other children than different!
- Importance of inclusive experiences/activities
- No one can predict the future (this is true for ANY child!)
Why have a diagnosis?

- gives parents explanation for features
- enables preventative medical care
- up to date information/networking
- accurate genetic counseling
- option of prenatal diagnosis if available
- qualifies child for certain services
- helps teachers design appropriate curriculum
Biology Review

DNA Molecules

Cell

Nucleus

Chromosomes
Achondroplasia

- Autosomal dominant
- 80% new mutations
- Adult height ~3-4 ft
- Shortened proximal extremities, frontal bossing
- Normal intelligence
Cleidocranial dysostosis

- AD inheritance
- Absent clavicles
- Delayed closure of fontanelle
- Dental prods
Fragile X Syndrome

- X-linked inheritance; shows imprinting effects
- Long face with large ears; hyperextensible joints, large testicles in puberty
- Most common inherited type of MR
- Autistic features in some
Down Syndrome

- Most trisomy 21; small % translocation
- Upslanted palpebral fissures, epicanthal folds, small ears, single palmar crease
- All features seen in general population as well – it is the PATTERN that raises suspicion of diagnosis
- 50% with heart probs
- Lifespan 50-60 years
Down Syndrome
<table>
<thead>
<tr>
<th>Maternal Age</th>
<th>Risk of Down Syndrome</th>
<th>Total Risk for Chromosome Abnormalities</th>
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<tbody>
<tr>
<td>20</td>
<td>1 in 1667</td>
<td>1 in 526</td>
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<td>25</td>
<td>1 in 1250</td>
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<td>1 in 952</td>
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### Down Syndrome Development Compared to "Normal" Development

<table>
<thead>
<tr>
<th></th>
<th>Children With Down Syndrome</th>
<th>Normal Children</th>
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<tbody>
<tr>
<td></td>
<td>average</td>
<td>range</td>
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<tr>
<td>smiling</td>
<td>2 months</td>
<td>1½ to 4 months</td>
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<tr>
<td>rolling over</td>
<td>8 months</td>
<td>4 to 22 months</td>
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<tr>
<td>sitting alone</td>
<td>10 months</td>
<td>6 to 28 months</td>
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<tr>
<td>crawling</td>
<td>12 months</td>
<td>7 to 21 months</td>
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<tr>
<td>creeping</td>
<td>15 months</td>
<td>9 to 27 months</td>
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<tr>
<td>standing</td>
<td>20 months</td>
<td>11 to 42 months</td>
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<tr>
<td>walking</td>
<td>24 months</td>
<td>12 to 65 months</td>
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<tr>
<td>talking, words</td>
<td>16 months</td>
<td>9 to 31 months</td>
</tr>
<tr>
<td>talking, sentences</td>
<td>28 months</td>
<td>18 to 96 months</td>
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</tbody>
</table>

(Pueschel, 1978)
Turner Syndrome

- Girls with one X chromosome rather than two
- Small stature
- Heart/kidney probs
- Infertility/ovarian probs
- Normal intelligence
Bardet-Biedl syndrome

- Features: obesity, retinitis pigmentosa, polydactyly, hypogonadism, MR
- AR inheritance
Prader-Willi Syndrome

- Hypotonia in infancy
- Small stature, obesity, hypogonadism, MR
- FTT followed by overeating behavior
- Features overlap other syndromes as well – be certain of diagnosis as it may change your rr!
- Chromosome 15 deletion – paternal (low rr)
Angelman Syndrome

- Happy disposition
- Wide mouth with widely spaced teeth
- Seizures
- Ataxia
- MR/absent speech
- Chromosome 15 deletion – maternal
Cleft Lip/Palate

- Multifactorial inheritance
- Can involve lip alone or lip and palate
- Can be bilateral or unilateral
- Can be associated with many syndromes; risk depends on cause
Spina Bifida

- Multifactorial inheritance
- 1/500 pregnancies affected; all women screened by AFP and most with ultrasound
- 80% also with hydrocephaly
- Risk can be reduced by folate supplementation
Fetal Alcohol Syndrome

- Poor growth, microcephaly, seizures, smooth philtrum, thin upper lip, MR
- Due to prenatal exposure to alcohol
- Unclear what is threshold for effects
- One of few causes of MR that is entirely preventable!
Why I like genetic counseling...

- Always learning new things as the field is changing rapidly!
- Opportunity to develop relationships with families and children over the years
Most Unusual Cases!!

- Trimethylaminuria ("fish odor syndrome")
- Kabuki make-up syndrome
- Oto-palato-digital II syndrome family
- Albinism
- Long QT syndrome
- Smith-Magenis child of first cousins
A child is like a butterfly in the wind
Some can fly higher than others;
But each one flies the best it can.
Why compare one against the other?
Each one is Different.
Each one is Special.
Each one is Beautiful.