Genetic Conditions and Services: An Introduction

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Goals

• Determine which children/families may benefit from a Genetics evaluation

• Explain to families the possible benefits and limitations of a Genetics clinic visit

• Explain to families what to expect at a Genetics clinic visit
WHAT IS A GENETIC CONDITION AND WHEN TO CONSIDER REFERRING TO GENETICS?
Genetic Condition

• Difference in a gene or chromosome that causes disease.
  – May be inherited from a parent or parents.
OR
  – May be new to the child, not inherited from parents.
Chromosomes

- Human cells contain 23 pairs of chromosomes (total = 46 chromosomes)
  - 1-22, X and Y
  - One chromosome of each pair inherited from each parent
Example of a Genetic Condition

- Chromosomal abnormality
  - Down syndrome

- Examples of other chromosomal abnormalities
  - Trisomy 13
  - 22q11.2 deletion syndrome
Genes

• Each chromosome contains many genes.

• Genes are sequences of bases that encode instructions on how to make proteins.
  – Similar to recipes.

• Each gene has a unique DNA sequence.
Genes

• Humans have ~20,000 genes.

• Gene sequence
  – Made up of 4 nucleotides
    • A, G, C, T
  – Change in the sequence is called a “variant.”
A Gene Is Similar To A Recipe

Deletion

The pkg has red dots.
The pkg has red dogs.

Sequence variant
Example of a Genetic Condition

• Single Gene Disorder
  – Duchenne Muscular Dystrophy
  – Noonan syndrome
  – Fragile X syndrome
  – Thousands of other single gene disorders
When to Consider Referring to Genetics

• **Developmental delay**
  – Unexplained global developmental delay
  – Autism
  – Unusual facial features accompanied by developmental delay

• **Abnormal growth and development**
  – Failure to thrive
  – Short stature
  – Overgrowth
When to Consider Referring to Genetics

- **Congenital anomalies**
- **Neurological abnormalities**
  - Congenital hypotonia or hypertonia
  - Developmental regression
  - Progressive muscle weakness
  - Intractable seizures
  - Abnormal brain MRI
When to Consider Referring to Genetics

- Congenital eye defects/blindness
- Significant hearing loss
  - Not related to chronic ear infections
- Unusual skin findings
  - Numerous hyper- or hypo-pigmented skin lesions
  - Albinism
- Family history
BENEFITS AND LIMITATIONS OF A GENETICS CLINIC VISIT
Family Example: Importance of Diagnosis

- 2 year old female, who we will call Emily
- Family is Spanish-speaking. The parents sought out community Intake and Referral Services.
- Emily receives in-home Occupational, Physical, and Speech Therapy.
- Emily is able to sit unsupported, is not able to pull to a stand, babbles, has no words, and hand movements are not purposeful.
- Mother notes that Emily’s functioning is getting worse despite receiving multiple therapies.
Feelings expressed by mom during home visit

- Frustration
- Anger
- Guilt
- Blame
The Family’s Concerns

• Because Emily continues to lose skills, mom thinks the therapists and doctors may not be providing appropriate services. She and her husband have discussed moving, as they think better services may be provided elsewhere. They will do ANYTHING to help their child.

• Emily is crying and frequently inconsolable. Parents think the crying started after a surgery. They wonder if Emily is in pain. They wonder if their choice to proceed with surgery has caused her to be in pain/inconsolable.

• Emily was scheduled and waiting to be seen by Genetics Clinic.
Benefits and Limitations

• What does this child and family have to gain by going to genetics clinic?

• What are the possible limitations for which the family should be prepared?
Follow Up

Phone call from the mother one year later with an update and a diagnosis.

- Emily was diagnosed with Rett syndrome.
Rett Syndrome

- Typically normal development during the first 6 to 18 months of life
- Then, rapid regression in language and motor skills, followed by stabilization
- Loss of purposeful hand use / Repetitive hand movements
- Screaming fits and inconsolable crying by age 18 to 24 months
- Autistic features
- Seizures
- Acquired microcephaly, poor growth, feeding difficulties
- Gait ataxia
The Family’s Reaction

• Now we understand why she was losing skills.
  – We know we aren’t doing something wrong and neither are the providers.
  – Relief
  – We can move forward because we know what to expect.
Genetics Clinic – Benefits to This Family?

• Help the parents and providers understand what to expect.
  – Provide appropriate developmental guidance.

• *Relieve blame/guilt.

• Help providers manage other possible health issues (seizures, scoliosis?).

• Help providers search for literature regarding appropriate therapies and surveillance/appropriate goals.

• Connect the family to research studies/clinical trials (if interested).

• Connect the family to other families and children with Rett syndrome.
  – Rettsyndrome.org
Another Benefit? – Inheritance

- Rett syndrome is caused by a disease-associated variant in the MECP2 gene located on the X chromosome
  - Females present with Rett syndrome.
  - A disease causing variant in a male would typically result in miscarriage or stillbirth.
- ~99.5% of cases are single occurrences in the family.
  - Mother may be offered testing for genetic variant identified in child.
Genetics Clinic – Limitations for This Family

• No cure available
GENETICS CLINIC VISIT

What to Expect
Before the Genetics Clinic Visit

• Referral (Please specify language of family in referral.)
  – Physician will be contacted for patient records, if not provided in referral.
  – Family may receive phone call if more information is needed.

• Wait List
  – From 1 month to 10 months

• Appointment
When family arrives for clinic visit

• Family checks in for clinic appointment.
• Care Assistant will obtain height and weight, medication list, etc.
• Appointment may last from ~45 minutes to one hour.
What happens during a Genetics Clinic visit?

• Genetic Counselor typically comes into room next.
  – Determines what the family hopes to gain from visit
  – Child’s Medical History
  – Family History
  • 3 Generation Pedigree
Pedigree

• Health and developmental information on 3 generations
  – Siblings, parents, aunts, uncles, first cousins, grandparents
• Ancestry
• Consanguinity
What happens during a Genetics Clinic visit?

Genetic Counselor and Pediatric Geneticist

- Physical Exam
  - Detailed examination

- Discussion of available testing/diagnostic considerations

- If testing is ordered:
  - Family goes to lab for blood draw after clinic visit.
  - Insurance preauthorization
Genetic Testing Results

• If a diagnosis is identified, family is often asked to return to clinic for a Genetic Counseling appointment.
  – Information about condition
    • Medical surveillance
    • Common features of condition, what to expect
    • Support groups for this condition?
  – Information about inheritance of the condition.
    • Other family members who may benefit from testing.
Sometimes Genetic Testing Occurs without a Pediatric Genetics Clinic visit

– Prenatal genetic testing
  • Prenatal Genetic Counseling
  • Confirmatory testing/Genetics Follow-up may be performed after birth

– Testing immediately after birth
  • NICU
  • Newborn nursery
  • A geneticist/genetic counselor may see the child/family during an inpatient stay.

– Due to shortage of genetics providers, pediatricians or specialty providers may order genetic testing
Resources

• Genetic Counselors
  – [www.nsgc.org](http://www.nsgc.org) Find a Genetic Counselor

• GeneReviews
  – Overview of information about specific genetic conditions.
    • Then search for specific condition.