The Heartland Regional Genetics Network
Overall Objective

• Expand capacity to improve access to pediatric, metabolic, and adult genetics services, targeting medically underserved populations that are defined by disparities in geography, socio-economic status, and race/ethnic groups, in the Heartland Region
Regional Genetics Networks
Heartland Regional Genetics Network

Heartland serves the following eight states: Arkansas, Iowa, Kansas, Missouri, Nebraska, North Dakota, Oklahoma, and South Dakota.

Coordinating Center: University of Arkansas for Medical Sciences
Heartland’s Hispanic Access Project

- Heartland has coordinated many projects
- Hispanic Access Project
  - Genetic Service providers noticed that they were serving fewer than expected Hispanic children in their clinics and some waited too long before being seen.
  - Designed and implemented a qualitative research project by Interviewing families from the Hispanic communities who are caregivers of a child with a genetic condition.
    - Interviewed 26 Spanish speaking families in Oklahoma, Kansas and Nebraska
    - Analyzed the data
    - Results and recommendations (March 2015)
What are the barriers to accessing genetic care?

1. Genetic Services (no or delayed referral)
2. Families perception of Genetic Services
3. Information Delivery
4. Families Expectations of Medical Care
5. Language barrier and translation Service
6. Cultural Barriers
7. Community programs (Facilitators)
Two Observations regarding community (facilitators)

- Families learn about genetic services and other services though the Hispanic (bilingual coordinators) from the Parent to Parent organizations.
  - Families form strong and trusting relationships with the Hispanic coordinators.
- Families many times are referred to early intervention services (or other services) before being referred to genetic services.
  - Families form strong and trusting relationships with the home visiting staff and the bilingual interpreters.
  - Many of the families asked the EI provider about their child's genetic condition.
Hispanic Access Advisory Committee (HAAC)

Members:

Parents
Parent to Parent coordinators
Early Intervention
Genetic Alliance
Genetic Service Providers
Heartland Regional Genetics Network

GOAL:
Create new and enhanced pathways to genetics services

- Strategy #1: Practice model
- Strategy #2: New Clinics
  - Outreach
  - Telehealth
- Strategy #3: Education
  - ECHO model
  - Home visitors
  - Family support organizations
- Strategy #4: All partners comprising a learning community supported by Heartland
Heartland Regional Genetics Network

Partnership Trios

- **Goal:** Increase access to genetics services for underserved populations despite shortage of genetics professionals
  - new / enhanced pathways
  - increasing competency of primary care providers
  - increasing awareness, knowledge, support in primary care, family support, and public health

- **Methodology:**
  - practice facilitation / QI strategies
  - building partnerships
  - education (specific groups; annual meeting; telemed skills)
  - existing public health programs
  - telemedicine
Genetic Services in the State of Kansas

• Wichita
• Kansas City (KS and MO)
• Hays (planning phase)
• Other sites in Kansas?
• KU Wichita Genetics Clinic
  • Wichita, KS

• Prenatal and Pediatric Genetics

• Developmental delay, birth defects, seizures, autism

• Innovative delivery – extending genetic service by telemedicine

• Phone: 316-962-2153
Children’s Mercy - Genetics

• Pediatric Genetics
  – Genetics and Dysmorphology Clinic
  – Metabolic Genetics Clinic

• To make an appointment
  – Phone: 816-234-3771
Genetic Conditions and Services: An Introduction

Jennifer Roberts, MC, MS, CGC
Laboratory Genetics Counselor
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**
How to Approach Families about Genetics Services

• Start with what they already know about their child (e.g. developmental delay)

• ASK if they would like to know more about what is going on? If yes…

• There are medical specialists that can help to figure out:
  – What is causing it?
  – What we can do about it?
  – What may the future hold?
  – Could it happen again?
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

![Pedigree Diagram]

*Children’s Mercy*
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

• Heartland Regional Genetics Network
  – https://www.heartlandcollaborative.org/

• Genetics Education Materials for School Success
  – https://www.gemssforschools.org/

• National Society of Genetic Counselors
  – www.nsgc.org Find a Genetic Counselor