Creative Strategies to Access Genetic Care Across Genetic Counseling Disciplines

Moutain States Regional Genetics Network Genetics Summit

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Disclosures

Lauren Westerfield has no financial disclosures

Emily Magness has no financial disclosures

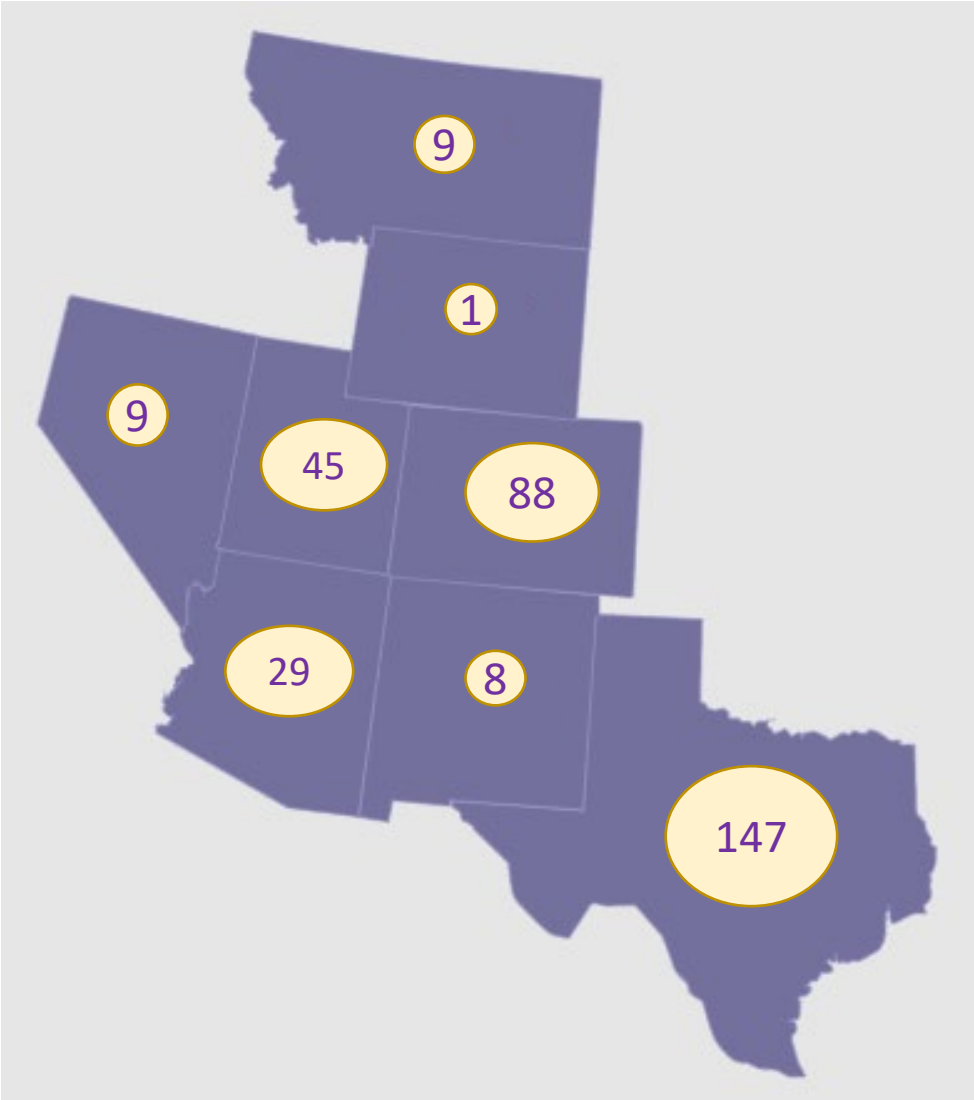
Learning Objectives

Identify gaps that can prevent or delay diagnostic workup for maternal and pediatric genetic diseases

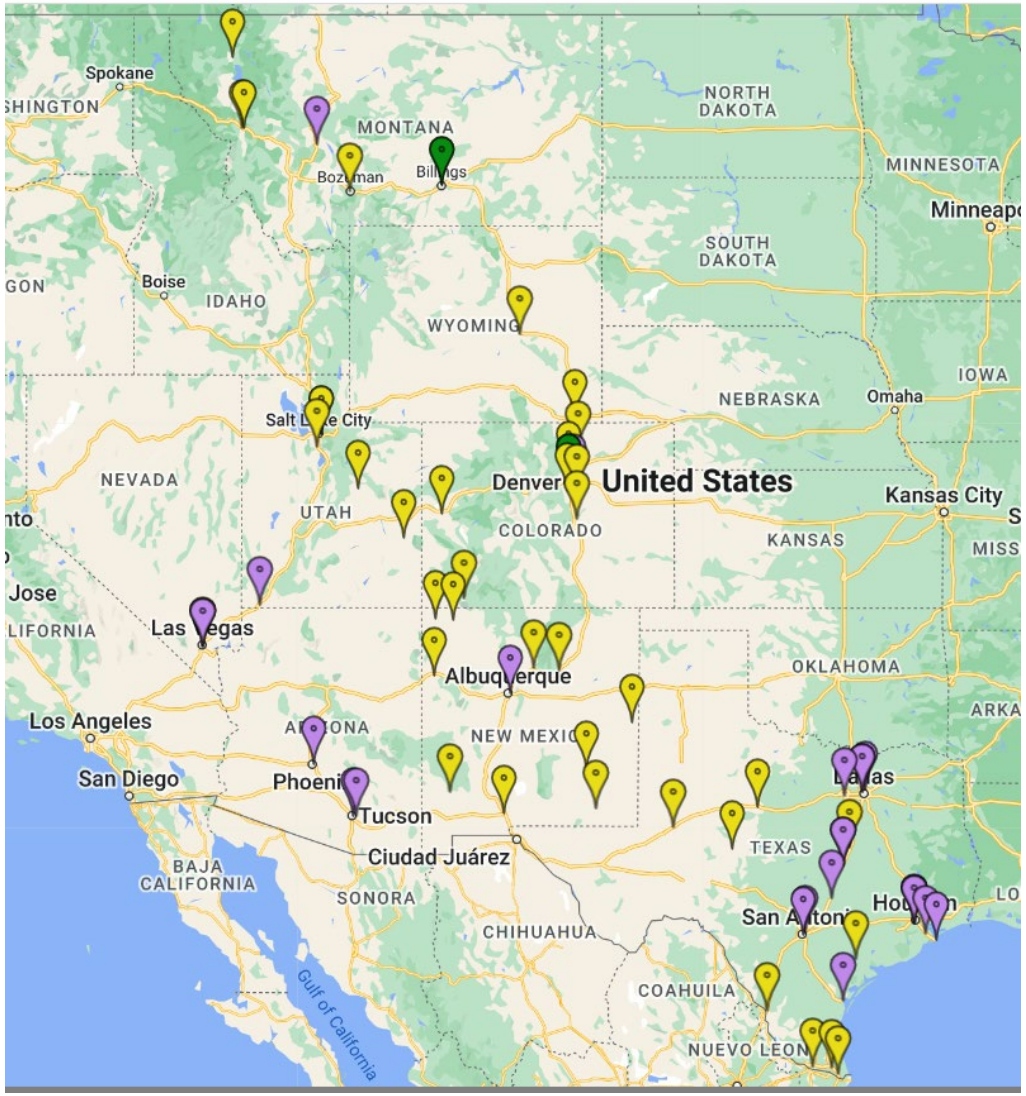
Demonstrate strategies for genetic counselors and medical professionals to address these gaps and facilitate improved medical management of these patients

Where do patients see a genetics provider?

Genetic counselor distribution

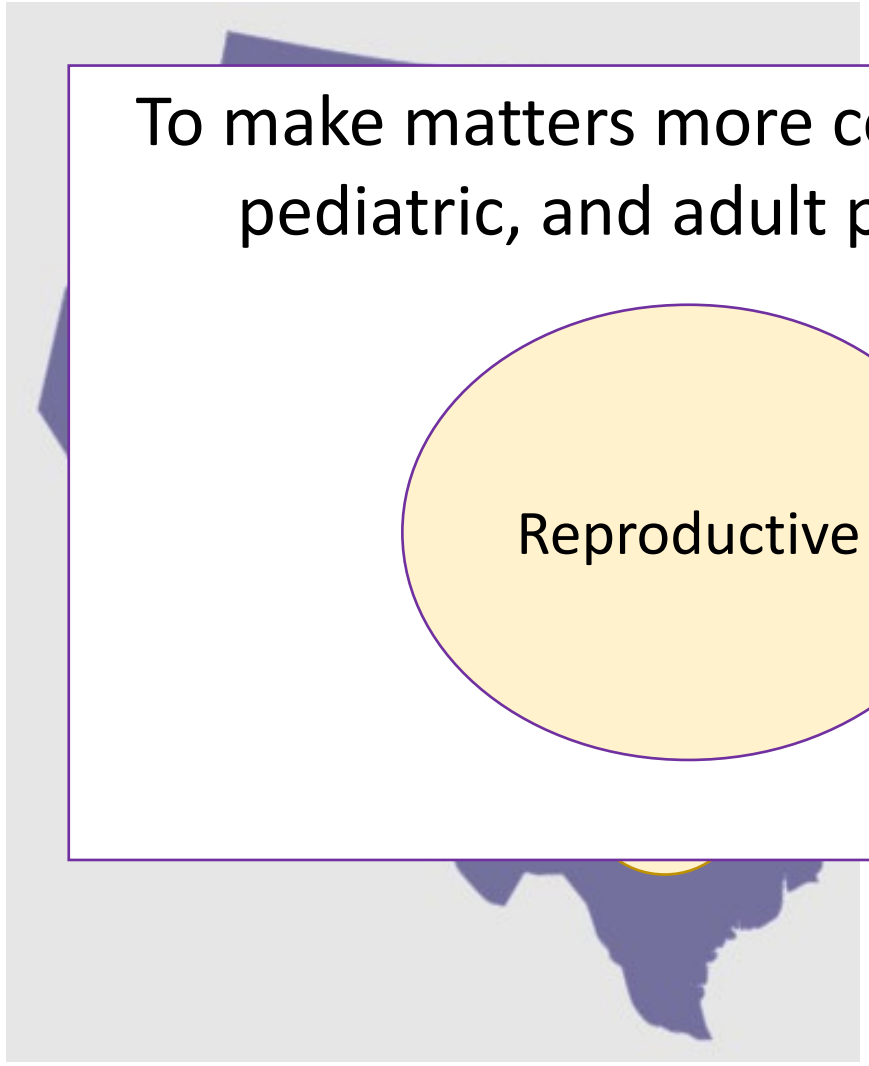


Genetic services distribution

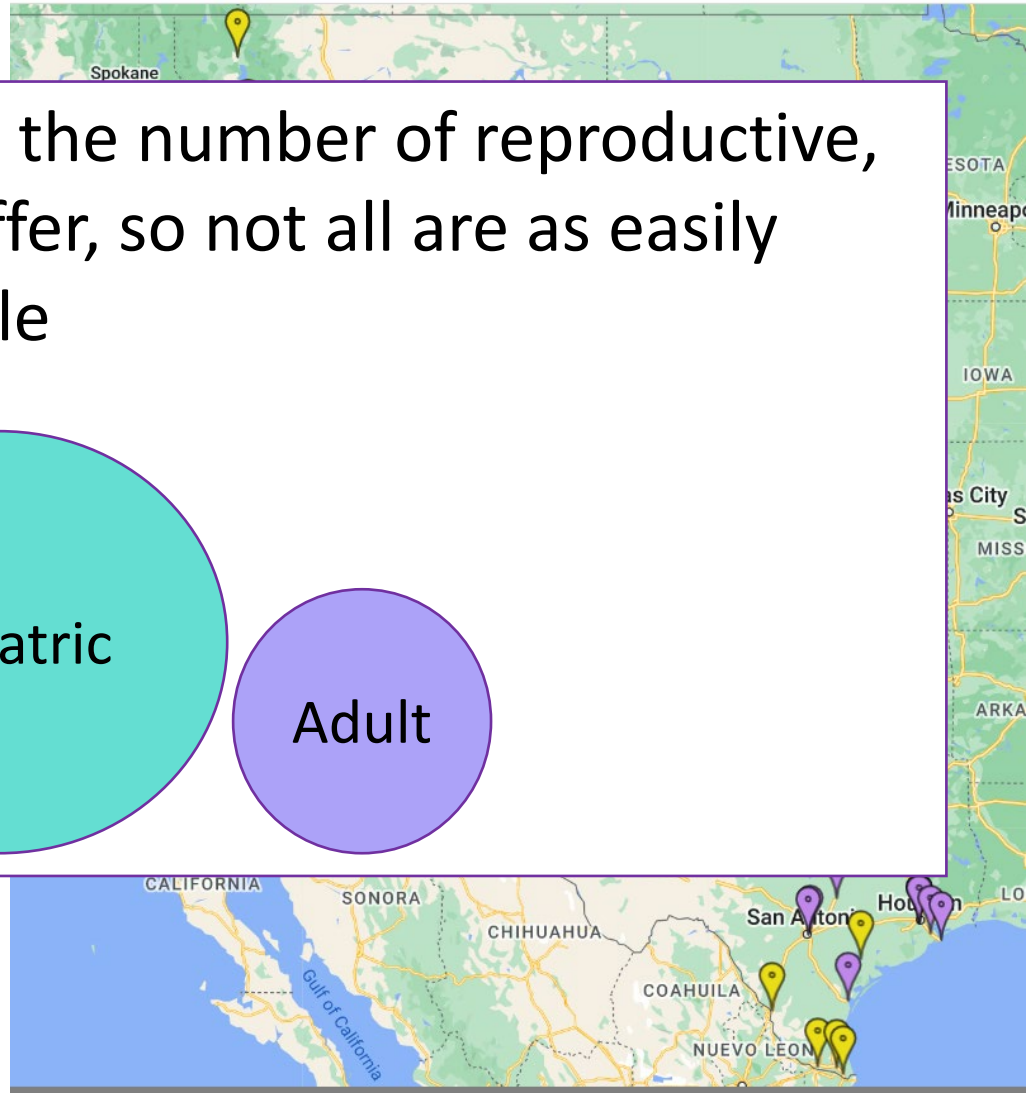


Where do patients see a genetics provider?

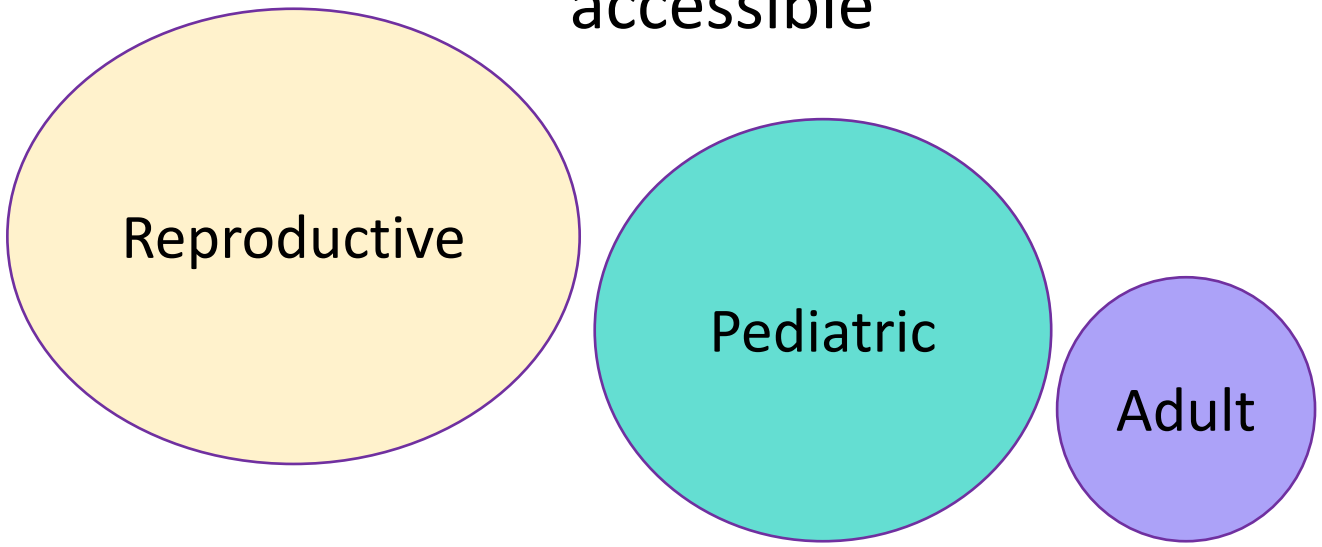
Genetic counselor distribution



Genetic services distribution



To make matters more complicated, the number of reproductive, pediatric, and adult providers differ, so not all are as easily accessible



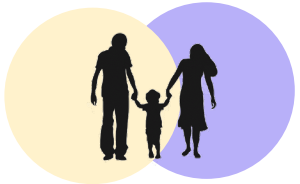
When do patients typically see a genetics provider?

Prenatal/Preconception

Newborn

Childhood

Teenager



Young Adult

Adult

Gaps in care

Prenatal

Limited prenatal care

Preconception

Lack of referral

Newborn

Testing completion

Childhood

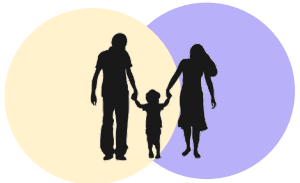
Loss of insurance

Teenager

Lost to follow up

Self-management

Young Adult

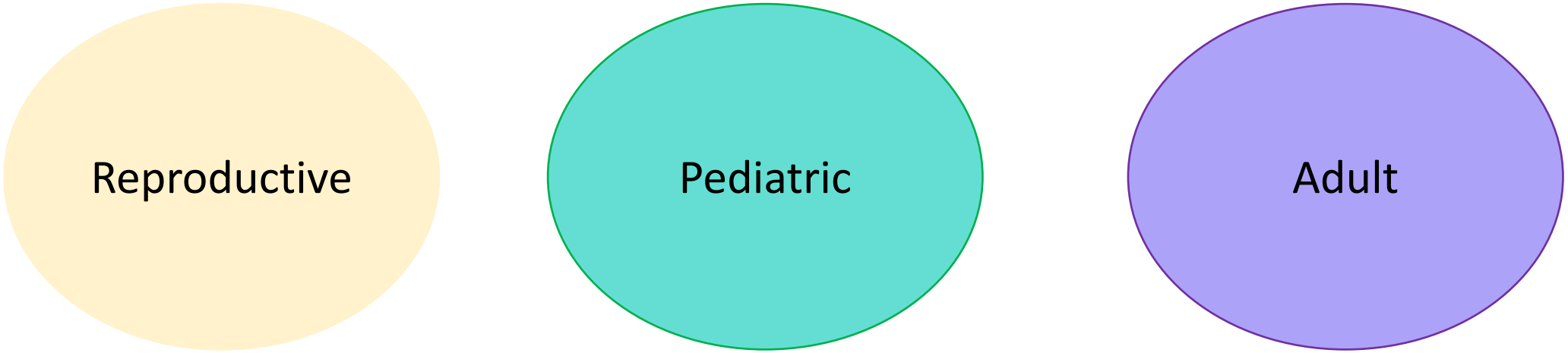


Provider accessibility

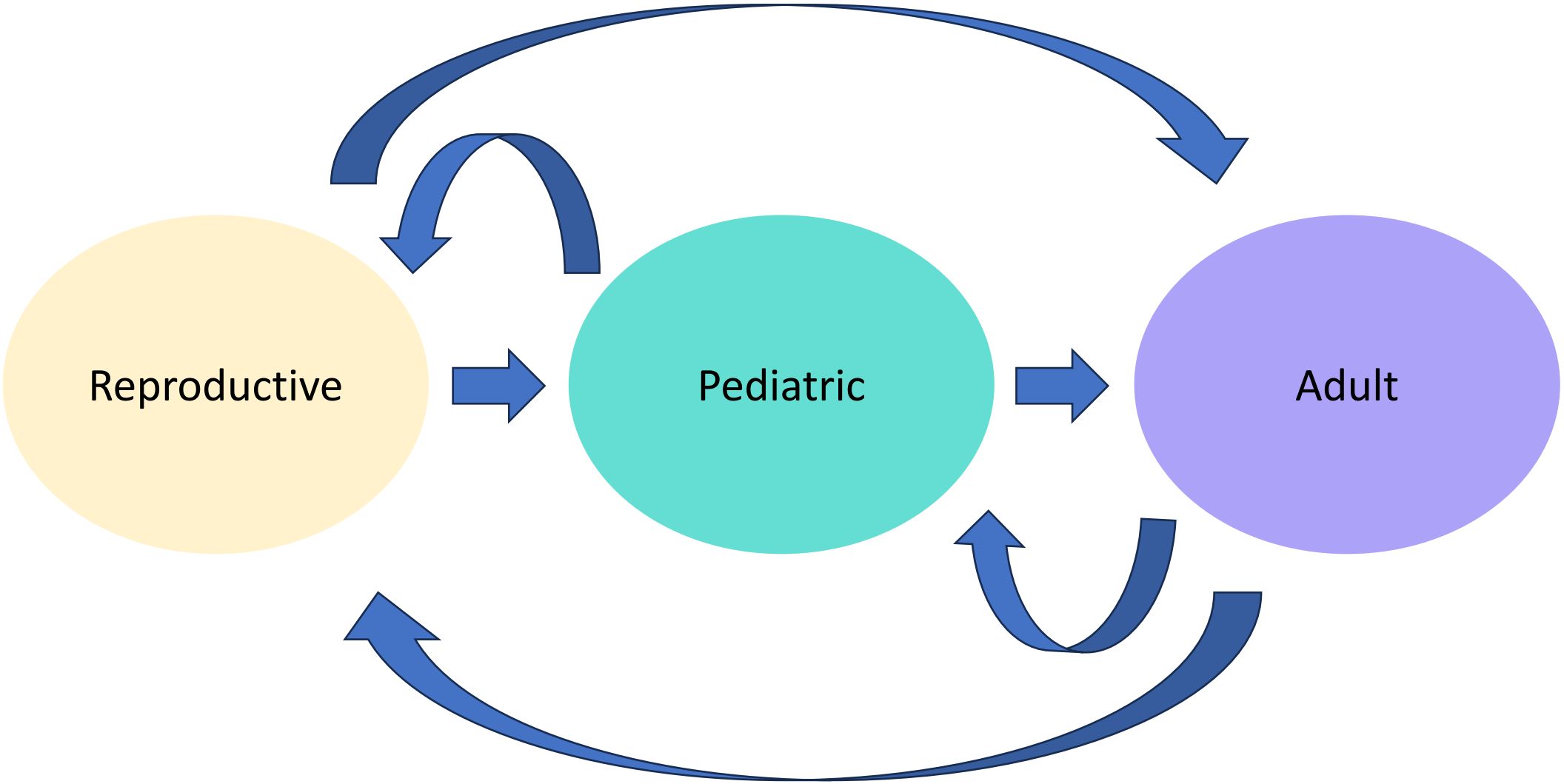
Adult

Genetics in Healthcare

- Healthcare is fragmented between age groups and "life stages"



- Genetic disease is multigenerational and impacts patients differently across their lifespan


Reproductive
Pediatric
Adult

Genetics in Healthcare

- Transitions between "life stage" are difficult
  - 2018 study found that 83% of youth with special healthcare needs and 86% of children without special healthcare needs were not receiving transition medicine support
- Barriers to transitions are plentiful
    - Systemic differences between pediatric and adult care
    - Cost prohibitive and insurance coverage
    - Lack of providers

What can genetic providers do to bridge care gaps, within the constraints of the healthcare system?

Practice Background and Scope

Genetic counselors in Texas do not:

- Have licensure and cannot bill independently for services
- Order genetic testing or make medical recommendations without physician supervision
- Conduct physical examinations or order other accessory biochemical or imaging studies relevant to the patient workup

We work at a large tertiary care center with level IV NICU capabilities and multiple geneticists and genetic counselors across all life stages

Our experiences and resources may not reflect those of individuals working in other centers, states, or medical practices

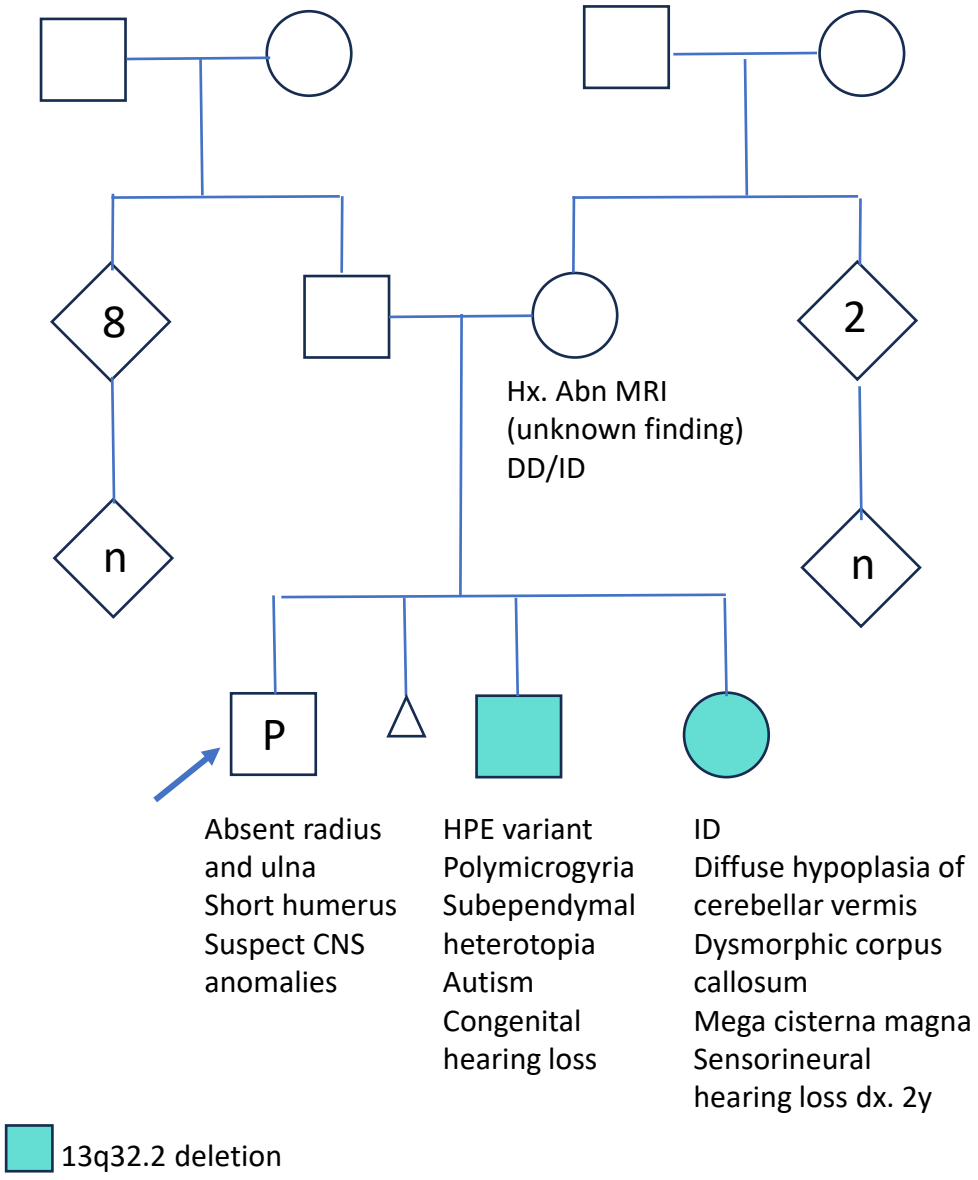
Case Series: strategies to access genetic care across life stages

Case example: Prenatal

- Initial presentation to fetal center:
  - 37 y.o. G4P2012 at 22w1d, fetus with absent right radius and ulna, short humerus, and concern for CNS anomalies.
  - Imaging severely limited by body habitus, but some views looked concerning for monoventricle
- Concerns about patient's cognitive capacity during counseling session
- Significant family history disclosed during session

Family history

- Concerns:
  - shared genetic etiology in family
  - suspect mild intellectual disability in the mother
  - Most likely AD or possibly XL
- With permission, reviewed the EMR for her other two children and confirmed they both have a 1.710 Mb deletion of 13q32.2q32.3 which includes *ZIC2* (one of the holoprosencephaly genes)



Gaps to address

- "Noncompliance" versus parental reduced capacity due to own genetic diagnosis
- No prior parental testing
- Mother unable to recall her children's diagnosis or genetic mutation

Strategy

- Parental testing
  - Mother has pregnancy Medicaid now, start with testing her. FOB uninsured but also less likely to carry the deletion based on reported history.
  - In person visits, visits timed with visits for her children at the same hospital, provider awareness of her diagnosis, involvement of social work
- Mother unable to recall her children's diagnosis or genetic mutation
- Noncompliance vs reduced cognitive capacity

Strategy

- Parental testing
- Mother unable to recall her children's diagnosis or genetic mutation
  - Integrated EMR allowed for easy retrieval of family testing results and phenotypic information
  - Written family letter and copy of results, emphasized importance of sharing this information with future medical providers
- Noncompliance vs reduced cognitive capacity

Strategy

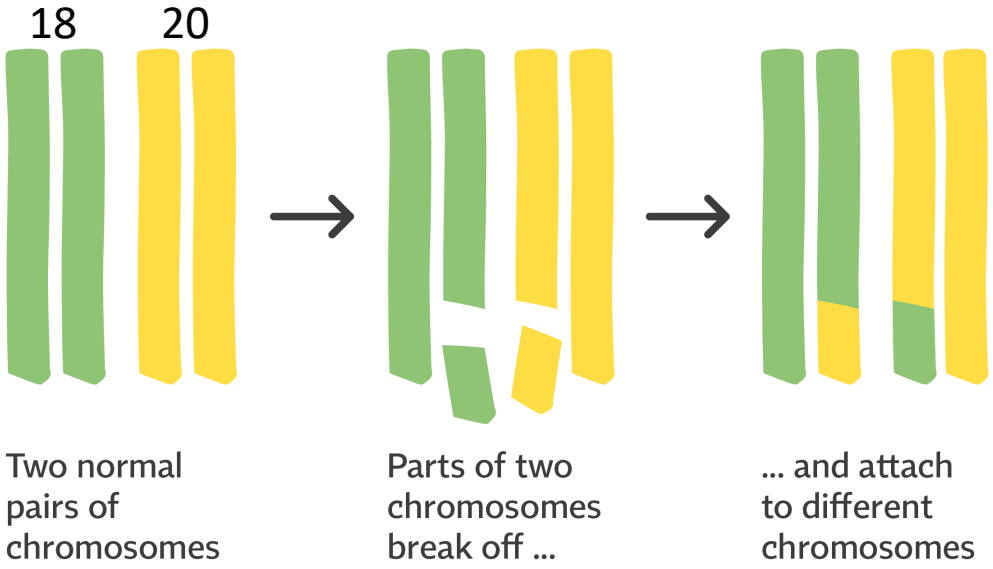
- Parental testing
- Mother unable to recall her children's diagnosis or genetic mutation
- Noncompliance vs reduced cognitive capacity
  - Telemedicine not a reliable option due to difficulty with following multi-step instruction and no home internet
  - Arranged in person visits in conjunction with visits at the hospital for her other children when possible
  - OB and pediatric provider education and involvement of social work to reduce stigma of "noncompliance" and increased support to improve follow through with medical care
- Personal diagnosis very validating for patient

Case Example: Pediatrics

- Initial presentation in pediatrics clinic:
  - Two siblings, age 4 and 6, with history of global developmental delay and dysmorphic features
- Both found to have an unbalanced translocation between chromosomes 18 and 20
- Parental testing revealed their mother carried balanced translocation

Chromosome Translocation Overview

- Balanced = The right number of chromosomes are present, but in different places
- Unbalanced = There are extra or missing pieces of chromosomes
- A person with a balanced translocation can have a child with an unbalanced translocation



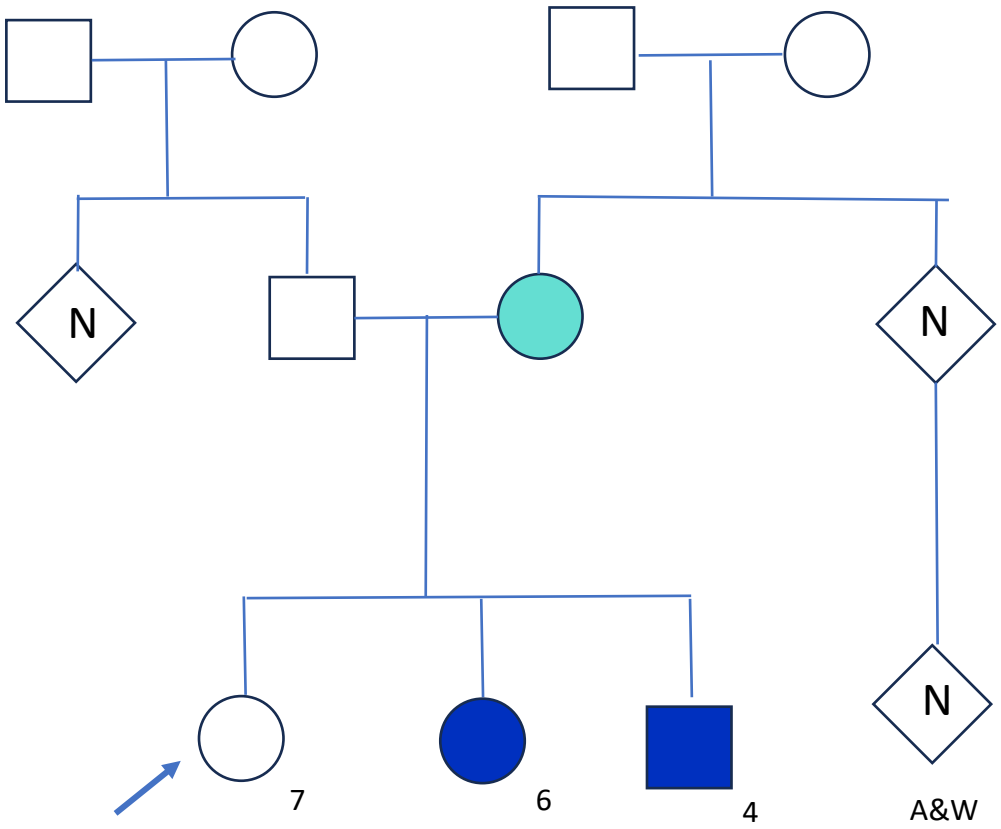
Balanced translocation

Unbalanced translocation

Extra piece of 20, missing piece of 18

Source of Graphics: Syndromes Without a Name UK

Family History



There is an older sister who is healthy and could be at risk of carrying the balanced translocation

Unbalanced 18;20 Translocation

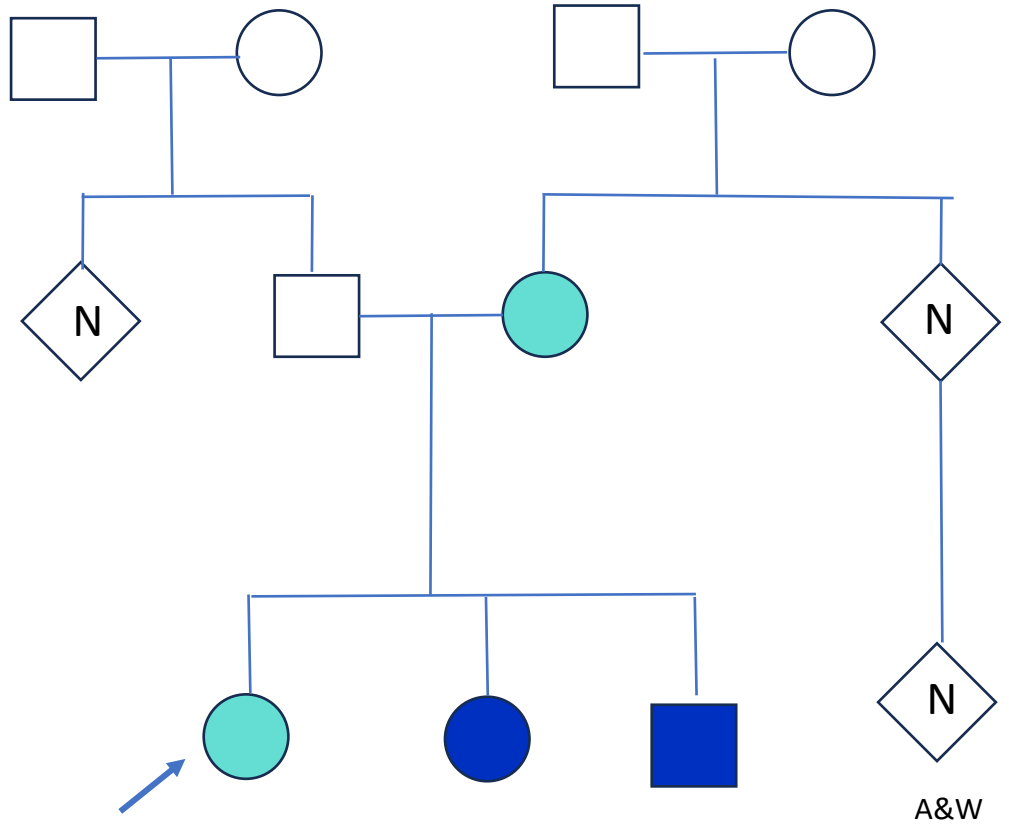
Balanced 18;20 Translocation Carrier

Fast forward 10 years…

- Siblings return to the Genetics clinic for routine follow up
- Their 17-year-old sister also attends appointment with siblings and mother
  - Sister has public health insurance as a minor and is at risk of no longer having health insurance after age 18 in Texas
  - Initial counseling is provided about risk of carrying the balanced translocation
  - Plan made to return when she is 18 to decide about genetic testing
    - Coordinated referral with her PCP

Follow up visit with now 18-year-old sister

- Test is ordered: She is a carrier of the balanced translocation
- Genetic counseling provided
  - Reproductive risks
  - How to find a prenatal genetic counselor



Gaps to Address

- Gaps to address
  - Teenager reproductive risks
    - Balance of preserving minor's autonomy for carrier testing vs. knowledge that teenagers can be sexually active
  - Transition period between pediatric and adult
    - Insurance coverage
    - Access to adult/reproductive genetics

Strategy

- Referral of teenagers to Genetics
  - Allows for repeat exposure to genetics concepts
  - Refer teenagers for reproductive counseling so they can have this discussion before they enter adulthood
- Include siblings and other family members in conversations and counseling
- Provide preconception counseling
- Outcome – This patient was able to establish her own care with genetics independently of siblings.

Strategy

- Referral of teenagers to Genetics
- Include siblings and other family members in conversations and counseling
  - Beat the clock—start conversations early before entering adulthood
- Provide preconception counseling
- Outcome – This patient was able to establish her own care with genetics independently of siblings.

Strategy

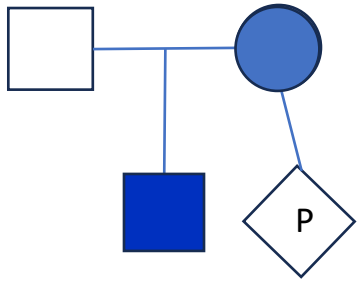
- Referral of teenagers to Genetics
- Include siblings and other family members in conversations and counseling
- Provide preconception counseling
  - Don't worry too much about specifics-- just that there may be something running in the family
    - Genetics notes often provide detailed information
- Outcome – This patient was able to establish her own care with genetics independently of siblings.

Strategy

- Referral of teenagers to Genetics
- Include siblings and other family members in conversations and counseling
- Provide preconception counseling
- Outcome: This patient was able to establish her own care with genetics independently of siblings.

Genetic diagnoses affect the whole family

- A woman's son is diagnosed with the metabolic disorder OTC deficiency
  - Metabolic X-linked condition, M>F
  - Female carriers can have life-threatening symptoms
- She discloses that she is 10 weeks pregnant and is referred to reproductive genetics
  - Confirmed carrier
  - She is now at risk to have a metabolic crash postpartum



Gaps addressed

- Multi-service care needed in a time sensitive manner
  - Utilized network contacts to facilitate referral, scheduling, and transfer of relevant records
- Rural home address distant to main service
- Highly specialized care team needed for safe delivery

Gaps addressed

- Multi-service care needed in a time sensitive manner
- Rural home address distant to main services
  - Able to use telemedicine for initial consults
  - Shipment of saliva collection kit from lab to patient home for carrier testing
- Highly specialized care team needed for safe delivery

Gaps addressed

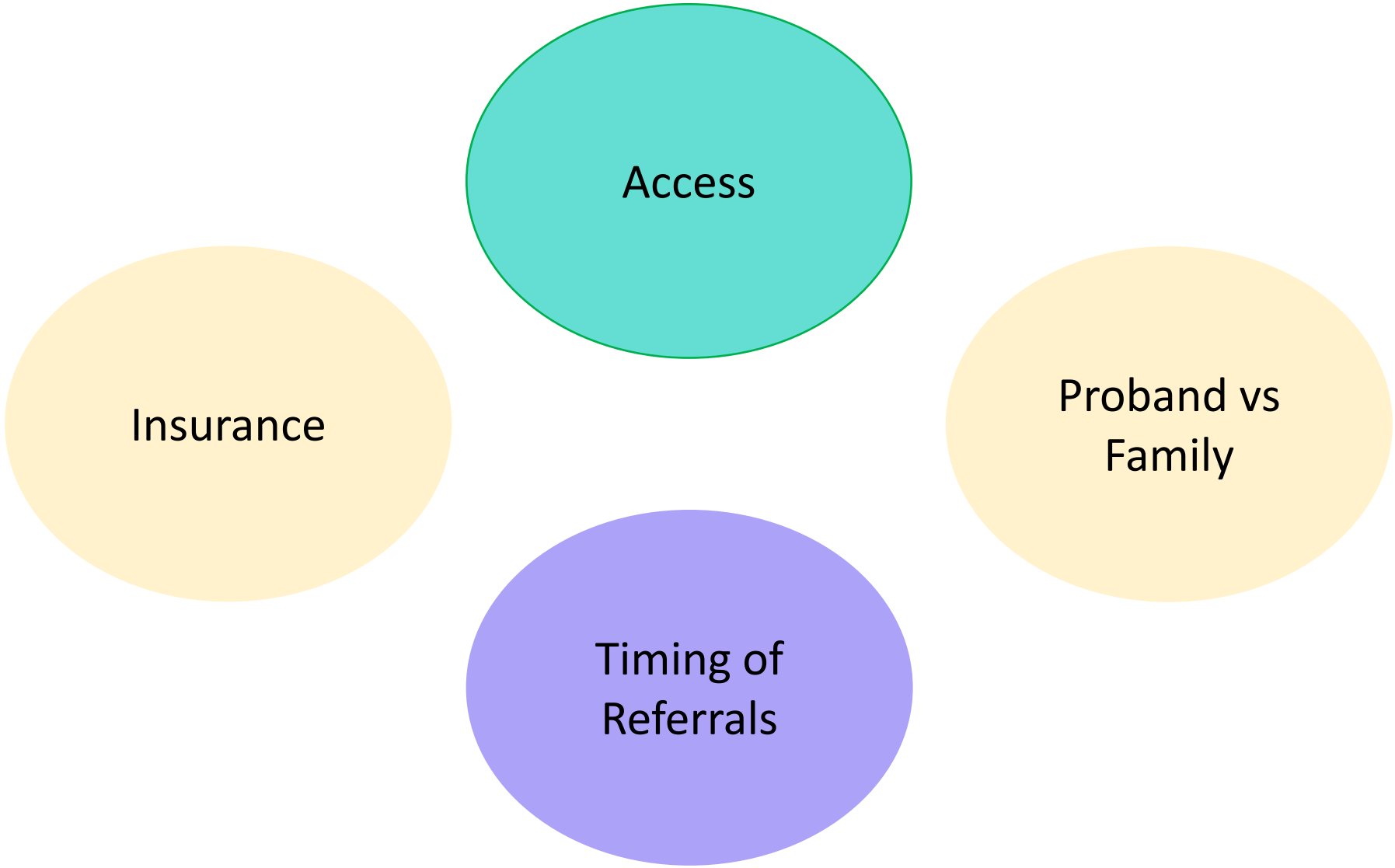
- Multi-service care needed in a time sensitive manner
- Rural home address distant to main services
- Highly specialized care team needed for safe delivery
  - She transfers care to an MFM and delivers at a hospital with an OTC delivery protocol. Mom starts recommended dietary modifications prenatally.

Gaps addressed

- Multi-service care needed in a time sensitive manner
- Rural home address distant to main services
- Highly specialized care team needed for safe delivery
- Outcome: Safe delivery, maternal postpartum symptoms caught quickly and metabolic crash avoided. Baby is tested after birth for OTC by pediatric genetics team

Wrap up

Common Themes



Other Strategies to Bridge Care Gaps

- Counseling letters for families and other healthcare providers about diagnosis
  - Shorter = better
  - Make contacts clear
  - Online resources/letters available on some genetic testing laboratories website, or alternative communication methods
  - Include copy of test results
- Utilize telemedicine and clinics with self-pay options
- Use your network
  - Ex: A prenatal counselor in your city might not be able to see your patient for their indication, but they likely know how to connect you to the right people

Other Strategies to Bridge Care Gaps

- Warm handoffs
- Be aware of sponsored testing programs, laboratory patient assistance plans, and research studies to facilitate testing and therapy options
- Procreative counseling for teens with genetic disorders
- Equal access for males/non-carrying partners
- Be aware your own biases and assumptions

In sum...

- Navigating a geographically, institutionally, and specialty fragmented healthcare landscape is difficult.
- Individuals with a genetic disorder and their families have the best outcomes when they have consistent care across their lifespan
- Look for creative solutions and develop networks and allies -- it takes a village
- You can't fix the system by yourself, but bridging the gap for one family could be life changing

Questions?

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