RAPID WHOLE GENOME SEQUENCING & PRECISION MEDICINE IN PEDIATRICS

MIAAP Annual Conference
Ann Arbor, MI
September 30, 2022
Disclosures

• No personal financial disclosures
• Non-financial disclosure
  • Institutional
    • Helen DeVos Children’s Hospital Foundation
    • Bronson Health Foundation
Goals

- Inspire passion for precision medicine in Pediatrics
- Michigan’s Project Baby Deer
- Family story
Why precision medicine in pediatrics?

• Improve health outcomes
  • Prevent long diagnostic odysseys
  • Optimize early treatments using genomic informed, personalized care
“Because primary care medicine combines the treatment of acute illness with disease prevention and anticipatory guidance, the primary care provider is in an ideal position to evaluate and treat patients for genetic disease.”

“The notion that genetic knowledge is only rarely needed will have to be replaced with a comprehensive approach that integrates “genetic thinking” into every patient encounter.”

“The ongoing provider–family relationship, coupled with the astounding number of advances in genetic and genomic testing, also necessitates a constant re-evaluation of past diagnosis or nondiagnosis.”


https://doi.org/10.1542/peds.2013-1032H
Impact of Genetic Disease

• Leading cause of morbidity and mortality in infants.
• There are over 3500 known monogenic diseases
  • Most of which present during the first 28 days of life.
• Rare diseases impact:
  • More people than cancer and AIDS combined
Chromosome to Gene to Protein

Cell

Chromosomes
Each chromosome is composed of one large continuous DNA molecule.

Gene
A gene is a segment of DNA that encodes a protein product.

Protein
A protein is a complex organic compound composed of hundreds or thousands of amino acids.

DNA

Nucleotides
- Adenine
- Thymine
- Guanine
- Cytosine

[Diagram of a cell with chromosomes, DNA, and nucleotides labeled]
## Sequencing Methodologies

<table>
<thead>
<tr>
<th>Methodology</th>
<th>Example</th>
<th>Look for errors in</th>
</tr>
</thead>
<tbody>
<tr>
<td>Single gene sequencing</td>
<td>The car was red. The car was red.</td>
<td>in a single sentence in the book</td>
</tr>
<tr>
<td>Targeted gene panel sequencing</td>
<td>The boat was blue. The boat was blue.</td>
<td>in a specific group of sentences in the book</td>
</tr>
<tr>
<td>Exome sequencing</td>
<td>The train was black. The ____ was black.</td>
<td>in the most important chapters in the book</td>
</tr>
<tr>
<td>Whole Genome sequencing</td>
<td></td>
<td>Look for errors in every single word in the book</td>
</tr>
</tbody>
</table>
Interim empirical treatment

Disease of Unknown Etiology
Search for etiological diagnosis
Genetic Testing 6 weeks
Treatment Modification
Improvement or worsening

• Discharged Home
• Palliative care
• Death
• Anxiety, Suffering
• Cost

Disease of Unknown Etiology
Search for etiological diagnosis
Genetic Testing 6 weeks
Treatment Modification
Improvement or worsening

• Discharged Home
• Palliative care
• Death
• Anxiety, Suffering
• Cost
~50% Disease of Unknown Etiology → Search for etiological diagnosis with rWG

Interim empirical treatment

35% Genetic Disease Diagnosis

25% Precision Medicine

20% improved outcomes Less cost and suffering

Genetic Testing 1.5–10 days

13

Courtesy: Vermont Oxford Network
- 130,000,000,000 Nucleotides sequenced
- 3,000,000,000 Nucleotides assigned
- 5,000,000 Nucleotide variants
- 750,000 DNA changes present in <1:100 people
- 1,000 DNA changes that could cause disease
- 1 Provisional diagnosis
What and Why?

An initiative created by a collaborative team from Michigan Health and Hospital Administration (MHA), pediatric clinical champions from across the Michigan, and Rady Children’s Institute for Genomic Medicine (RCIGM).

To introduce rWGS as a first-tier test for hospitalized infants and children with unexplained illness and suspicion of underlying genetic etiology.

To help neonatal and pediatric clinical teams provide fast, targeted, and life-saving, life-changing treatments.

To better understand barriers to testing access and work towards breaking those barriers down.
**Inclusion Criteria**

- Inpatient at a MI project site
- <18 years old
- Meets one of the following criteria:
  - Admitted to a critical care unit OR
  - Admitted to another high-acuity in-patient unit and is suspected of having a genetic diagnosis
- Meets one of the following criteria:
  - Within 1 week of admission OR
  - Within 1 week of development of an abnormal response to standard therapy for an underlying condition

**Exclusion Criteria**

- Patients whose clinical course is entirely explained by:
  - Infection or sepsis with normal response to therapy
  - Isolated prematurity
  - Isolated unconjugated hyperbilirubinemia
  - Hypoxic ischemic encephalopathy with clear precipitating event
  - Previously confirmed genetic diagnosis that explains the clinical condition (e.g. have a positive genetic test)
  - Isolated transient neonatal tachypnea
  - Trauma
  - Meconium aspiration
<table>
<thead>
<tr>
<th>PILOT SITE</th>
<th># OF CHILDREN WHO RECEIVED RWGS</th>
<th>CHILDREN DIAGNOSED (DIAGNOSTIC RATE)</th>
<th>CHILDREN WHOSE CARE WAS CHANGED (CHANGE IN MANAGEMENT RATE)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Beaumont Health – Dearborn</td>
<td>3</td>
<td>2 (67%)</td>
<td>2 (67%)</td>
</tr>
<tr>
<td>Beaumont Health – Royal Oak</td>
<td>6</td>
<td>0 (0%)</td>
<td>0 (0%)</td>
</tr>
<tr>
<td>Beaumont Health – Troy</td>
<td>1</td>
<td>1 (100%)</td>
<td>0 (0%)</td>
</tr>
<tr>
<td>Bronson Methodist Hospital</td>
<td>15</td>
<td>6 (40%)</td>
<td>7 (47%)</td>
</tr>
<tr>
<td>Children’s Hospital of Michigan</td>
<td>10</td>
<td>4 (40%)</td>
<td>1 (10%)</td>
</tr>
<tr>
<td>Helen DeVos Children’s Hospital</td>
<td>45</td>
<td>18 (40%)</td>
<td>11 (25%)</td>
</tr>
<tr>
<td>Sparrow Hospital</td>
<td>9</td>
<td>4 (44%)</td>
<td>3 (33%)</td>
</tr>
<tr>
<td><strong>TOTALS</strong></td>
<td><strong>89</strong></td>
<td><strong>35 (39%)</strong></td>
<td><strong>24 (27%)</strong></td>
</tr>
</tbody>
</table>

**PRELIMINARY RESULTS OF RAPID WHOLE GENOME SEQUENCING (RWGS):**

- 39% of rapid genomes resulted in a diagnosis confirmed by the treating physician
- 27% of children had changes to their care as a result of RWGS
- Oldest PBD patient: 17 years of age
- Youngest PBD patient: 1 day old
- Race: White - 72%; Black or African-American - 9%; Asian - 3%; Other - 9%; Unknown - 7%
- Ethnicity: Non-Hispanic - 43%; Hispanic - 11%; Other - 7%; Unknown - 39%
- Sex: Female - 36%; Male - 64%

**THESE CHANGES LED TO:**

- Avoided between 95 and 214 inpatient hospital days
- Multiple avoided surgeries and procedures (including lung biopsy, tracheostomy, muscle biopsy, and skin biopsy)
- Appropriate medications prescribed based on genetic diagnosis
- Initiation of a heart transplant

**PROJECT SAVINGS*: NET BENEFIT PER PATIENT DUE TO REDUCTION IN HOSPITALS DAYS AND MAJOR PROCEDURES**

$2,842

Per patient

**FOR A TOTAL OF**

$252,938

* as of 11/30/21

For more information, contact: keystone@mha.org
### CASES WITH ECONOMIC SAVINGS

<table>
<thead>
<tr>
<th>Site</th>
<th>Case</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Site 1</td>
<td>Case 1</td>
<td>Avoided skin biopsy</td>
</tr>
<tr>
<td>Site 2</td>
<td>Case 6</td>
<td>7-42 hospital days avoided</td>
</tr>
<tr>
<td></td>
<td>Case 11</td>
<td>7 hospital days avoided</td>
</tr>
<tr>
<td></td>
<td>Case 14</td>
<td>Avoided lung biopsy, Avoided transfer to another hospital for lung biopsy</td>
</tr>
<tr>
<td>Site 3</td>
<td>Case 1</td>
<td>Stopped medication, 2 hospital days avoided</td>
</tr>
<tr>
<td></td>
<td>Case 4</td>
<td>2 hospital days avoided</td>
</tr>
<tr>
<td></td>
<td>Case 5</td>
<td>Cancelled interventional radiology, cerebral angiogram; added medication</td>
</tr>
<tr>
<td></td>
<td>Case 10</td>
<td>Avoided muscle biopsy</td>
</tr>
<tr>
<td></td>
<td>Case 18</td>
<td>14-28 hospital days avoided</td>
</tr>
<tr>
<td></td>
<td>Case 19</td>
<td>14-28 hospital days avoided</td>
</tr>
<tr>
<td>Site 4</td>
<td>Case 2</td>
<td>Tracheostomy avoided, 28-84 hospital days avoided</td>
</tr>
<tr>
<td></td>
<td>Case 3</td>
<td>7 hospital days avoided</td>
</tr>
<tr>
<td></td>
<td>Case 4</td>
<td>14 hospital days avoided</td>
</tr>
</tbody>
</table>
Healthcare Professionals’ Attitudes toward Rapid Whole Genome Sequencing in Pediatric Acute Care

by Linda S. Franck 1,*, Andrea Scheurer-Monaghan 2,3, Caleb P. Bupp 4,5, Joseph D. Fehoum 3,6, Thomas J. Hoffmann 7, Manasi Deshpande 1, Madison Arechiga 8, and David P. Dimmock 8

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### Project Baby Deer: Health Professionals Survey - Results

#### Experience with rWGS

52% of respondents had been involved in the care of an inpatient infant or child for whom rWGS was ordered

24% of respondents had direct conversations with families about rWGS testing or diagnosed disorders

#### rWGS self-rated knowledge

“a little” (34%; n=103); “none” (29%; n=90)

64% of geneticists and genetic counselors reported “a lot” or “expert”

98% (n=124) of clinicians reported genetics education

53% (n=69) of direct care nurses reported any genetics education
## Factors influencing attitudes toward rWGS/Final linear regression model

<table>
<thead>
<tr>
<th>Term</th>
<th>Factor 1: Personal Capability</th>
<th>Factor 2: Potential/ Intention</th>
<th>Factor 3: Implementation</th>
<th>Total Attitudes Score</th>
</tr>
</thead>
<tbody>
<tr>
<td>Self-rated level of knowledge about rWGS</td>
<td>0.505 (0.415, 0.596)</td>
<td>0.44 (0.357, 0.523)</td>
<td>0.378 (0.273, 0.483)</td>
<td>5.62 (4.58, 6.66)</td>
</tr>
<tr>
<td></td>
<td>(&lt;0.001)</td>
<td>(&lt;0.001)</td>
<td>(&lt;0.001)</td>
<td>(&lt;0.001)</td>
</tr>
<tr>
<td>Clinical role (vs. non-clinical)</td>
<td></td>
<td>0.326 (0.114, 0.538)</td>
<td></td>
<td>3.7 (1.62, 5.78)</td>
</tr>
<tr>
<td></td>
<td></td>
<td>(0.0028)</td>
<td></td>
<td>(&lt;0.001)</td>
</tr>
<tr>
<td>Confidence about future insurance coverage for rWGS</td>
<td>0.219 (0.112, 0.326)</td>
<td>0.223 (0.119, 0.326)</td>
<td>0.208 (0.0928, 0.324)</td>
<td>3.01 (1.87, 4.15)</td>
</tr>
<tr>
<td></td>
<td>(&lt;0.001)</td>
<td>(&lt;0.001)</td>
<td>(&lt;0.001)</td>
<td>(&lt;0.001)</td>
</tr>
<tr>
<td>Concerns about potential long-term effects of genomic testing on patients/families</td>
<td>−0.266 (0.174, 0.358)</td>
<td></td>
<td></td>
<td>−1.95 (0.934, 2.97)</td>
</tr>
<tr>
<td></td>
<td>(&lt;0.001)</td>
<td></td>
<td></td>
<td>(&lt;0.001)</td>
</tr>
<tr>
<td>NICU/PICU (vs. other unit)</td>
<td>−0.285 (−0.464, −0.107)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>(0.0019)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Concern about racial disparities in use of genomic testing</td>
<td></td>
<td>−0.265 (0.167, 0.363)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>(&lt;0.001)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Site2 (ref = Site 1)</td>
<td></td>
<td></td>
<td>0.419 (0.216, 0.622)</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>(&lt;0.001)</td>
<td></td>
</tr>
<tr>
<td>Adjusted R²</td>
<td>0.43</td>
<td>0.44</td>
<td>0.32</td>
<td>0.51</td>
</tr>
</tbody>
</table>

Children 2022, 9, 357. [https://doi.org/10.3390/children9030357](https://doi.org/10.3390/children9030357)
<table>
<thead>
<tr>
<th>Barriers to rWGS adoption</th>
<th>Successes</th>
</tr>
</thead>
<tbody>
<tr>
<td>IRB—is this really QI?</td>
<td>Philanthropic support</td>
</tr>
<tr>
<td>Establishing new lab service agreements</td>
<td>Early engagement with MDHHS/Medicaid Policy</td>
</tr>
<tr>
<td>Hospital legal team approval</td>
<td>Telemedicine relationships with genetics</td>
</tr>
<tr>
<td>Economic modeling for rare disease</td>
<td>Favorable investment</td>
</tr>
<tr>
<td>New billing workflow for inpatient rWGS</td>
<td>Faster adoption of rWGS</td>
</tr>
<tr>
<td>Variable knowledge of/comfort with genetic testing</td>
<td>Improved subspecialty collaboration across centers</td>
</tr>
<tr>
<td>Key stakeholder turnover</td>
<td>Increasing interest in pediatric genomics</td>
</tr>
<tr>
<td>Competing responsibilities for project champions</td>
<td>Family champions for rare disease</td>
</tr>
</tbody>
</table>
Evelyn

Using rWGS, Evelyn was diagnosed very early on with KAT6b-OHDO syndrome. Her diagnoses allowed her physicians to research all symptoms and conditions related to her syndrome. They used this research as a guide to provide Evelyn with the best care plan suited to her needs. Without this genome project, Evelyn would not be home recovering and living her best life.

Kayde

Kayde was born with low muscle tone and was very floppy as an infant. Thanks to rWGS being available in the NICU, we were able to find out about his genetic difference. Since then, he has been set up with all the right specialists and therapies to help make him strong. This test could find more babies similar to Kayde and help provide answers for the best possible care. All babies and families should have access to quick answers and any early prevention.
Case

• 4 mo Amish M
  • born at 36wk GA (at birthing center with midwife but discharged home same day)
  • Prenatal care within community

• Admitted to hospital for “second opinion” at the request of the parents

• Two previous PICU admissions in Indiana Hospital
History

• Brief cyanotic episode after birth which reportedly (per parents) resolved when the midwife tried to stimulate suckling by placing formula in his mouth

• Always gets tired with feeding, no diaphoresis; parents note he has always had a weak cry, even since birth; also not as strong as other siblings (7th child)

• Seen at OSH ED about 6 weeks after birth for what was documented as bronchiolitis → discharged home

• About 10 days shy of 3 mo, admitted to OSH PICU after multiple reported episodes of apnea with cyanosis and marked desaturations to 36%
PICU Admission #1

- Initially intubated but extubated two days later
- Diagnosed with:
  - oropharyngeal dysphagia and started on thickened feeds
  - > 2cm umbilical hernia (reducible)
  - moderate L hydrocele
  - thickened spermatic cord on L side
  - L inguinal hernia
- Treated with ceftriaxone for pneumonia
- Discharged on hospital day 10 with PO thickened feeds, famotidine and close follow-up with surgery for hernia repair and consideration of G-tube
PICU Admission #1

- Diagnostics during admission:
  - Echo: PFO with L to R shunt present. Normal LV size and systolic function.
  - Swallow study: oral phase is abnormal; oropharyngeal dysphagia. Thin liquid with intermittent silent aspiration
• Re-admitted to same OSH six days after discharge because of multiple choking episodes, many of which occurred after feeds. No color change this time. He was also noted to be difficult to stimulate

• Day after admission, numerous episodes of apnea noted (unclear to what extent from documentation review) → placed on HFNC and NG feeding

• Treated for presumed PNA due to CXR infiltrate and fever

• Underwent laparoscopic Nissen, g-tube placement and umbilical hernia repair

• Continued to have apnea post-op, restarted on HFNC

• After diagnostics (next slide), started on caffeine for presumed central apnea.

• Able to be weaned off supplemental O2, tolerating feeds. Discharged on previous PO feeding regimen and miralax
PICU Admission #2

- Diagnostics during admission:
  - Newborn screen: confirmed to be normal. Important to note - collected 3 days before 3mo age
  - 24 hr EEG: no epileptiform activity
  - MRI Brain: within normal limits, including myelination patterns
Admission #3

- Admitted to our hospital within a couple weeks of most recent discharge (< 1 month from last admission). Hx 3d coughing, + ill sibling with cough.
- OSH ED day prior, dx with PNA and given dose of Azithro, parents did not continue outpatient due to limitations in obtaining.
- Fevers at home (Tmax 102°F)
- Day of admission - GM found him purple in the face and called EMS, episode lasted 1.5-5 min depending on parent asked.
- Told by EMS that he had sz en route to ED, given midazolam.
- Per EMS report, father was performing chest compressions on their arrival.
- Conflicting reports on what constituted sz activity suspicion and what prompted CPR.
Family and other pertinent Hx

• Unvaccinated
• 9yo sister requires hearing aids bilaterally
• Maternal second cousin “very similar” to pt, per parents. The child is now one-year old and “grew out of his issues.”
• Maternal uncle: seizures as a child, died at age 11 years (not febrile related)
• Maternal grandfather: seizures in childhood (not febrile related) - eventually resolved
• Developmental delay - does not sit with support
• Mild to moderate protein calorie malnutrition
• Hair has been falling out in clumps
Physical Exam

- Pertinent positives / negatives (on admission)
  - Weak cry
  - Exophthalmos bilaterally with mild yellow eye drainage bilaterally
  - Intermittent nasal flaring with moderate subcostal retractions and diffuse rhonchi
  - RLL crackles
  - G-tube OK
  - Pulses equally strong and present bilaterally
  - Healing surgical sites/scars

- Additional findings day after admission (my exam)
  - L frontoparietal alopecia (patchy)
  - Hypertelorism, pointed ears
  - L orbit protruding more than R
  - No murmur, though difficult to appreciate given HFNC and diffuse rales
  - Hypotonia globally, though mostly appreciated truncal
Remainder of admission

• + human metapneumovirus
• Transferred to PICU on night 2 of admission for increasing HFNC requirements
• Started on amoxicillin/clavulanic acid → CTX for concern for superimposed bact PNA
• Eventually weaned from HFNC → NC → RA
• > 24 hours no supplemental O2 prior to discharge
• Discharged 10 days after admission on:
  • Albuterol
  • caffeine (home med)
  • Polyethylene glycol (home med)
  • plans for outpatient OT, PCP follow-up
What I was thinking…

- Based on now three PICU admissions by age 4 months (all resp based with several episodes of cyanosis, apnea) + PE findings + family history + clinical course, I considered rWGS

- Geneticist in Indiana with specialty care of Amish population
  - Asked for photos (parents refused)
  - Asked me to encourage parents to submit samples (refused)

- Consult performed 3/11 (date of admission was 3/7 with PICU transfer on 3/9)

- Sample collected 3/11, received by lab 3/12 (Saturday)

- Patient discharged 3/16 (results still pending)

- Phone call from lab with prelim results discussed around 1 wk prior to result on 4/3 PM
**Test Results**

**TEST RESULT: PRIMARY FINDINGS IDENTIFIED, AND ADDITIONAL FINDINGS REPORTED**

*Note: Multiple regions of homozygosity (ROH) greater than 5 Mb were detected in this individual.*

### Copy Number Variants

<table>
<thead>
<tr>
<th>REPORT CATEGORY</th>
<th>VARIANT</th>
<th>CONDITION</th>
<th>SIZE</th>
<th>EVENT / ZYGOSITY (INHERITANCE)</th>
<th>VARIANT CLASSIFICATION</th>
</tr>
</thead>
<tbody>
<tr>
<td>VARIANTS RELATED TO PATIENT PHENOTYPE</td>
<td>chr9:140396866-140776781 del (9q34.3)</td>
<td>CHROMOSOME 9q34.3 DELETION SYNDROME</td>
<td>380 KB</td>
<td>Deletion / Heterozygous (Unknown)</td>
<td>Pathogenic</td>
</tr>
<tr>
<td>VARIANTS RELATED TO PATIENT PHENOTYPE</td>
<td>chr9:140785919-141018755 del (9q34.3)</td>
<td>CHROMOSOME 9q34.3 DELETION SYNDROME</td>
<td>233 KB</td>
<td>Deletion / Heterozygous (Unknown)</td>
<td>Uncertain significance</td>
</tr>
</tbody>
</table>

### Sequence Variants

<table>
<thead>
<tr>
<th>REPORT CATEGORY</th>
<th>GENE</th>
<th>VARIANT</th>
<th>CONDITION</th>
<th>ZYGOSITY (INHERITANCE)</th>
<th>VARIANT CLASSIFICATION</th>
</tr>
</thead>
<tbody>
<tr>
<td>VARIANTS POSSIBLY RELATED TO PATIENT PHENOTYPE</td>
<td>CARD11</td>
<td>c.1373T&gt;G p.Ile458Ser</td>
<td>CARD11-RELATED DISORDERS</td>
<td>Homozygous (Unknown)</td>
<td>Uncertain significance</td>
</tr>
<tr>
<td>VARIANTS POSSIBLY RELATED TO PATIENT PHENOTYPE</td>
<td>WDR19</td>
<td>c.3553A&gt;C p.Lys1185Gin</td>
<td>WDR19-RELATED DISORDERS</td>
<td>Homozygous (Unknown)</td>
<td>Uncertain significance</td>
</tr>
</tbody>
</table>

*Details on the variant(s) and gene(s) are located in the subsequent sections of the report*
Results

• Kleefstra Syndrome - Pathogenic variant
  • Multiple congenital malformation disorder characterized by moderate-severe intellectual disability with severe speech delay, childhood hypotonia, microcephaly, and facial dysmorphism.
  • Additionally there is a broad range of other associated anomalies including cardiac defects, genitourinary malformations, respiratory infections, epilepsy, neurological anomalies, obesity in childhood, autistic like features in childhood, and extreme apathy/catotonic-like features post puberty.
  • Of interest - neuromuscular disease? immune deficiency?
• Results given to family by phone on 4/4
• Genetics follow-up arranged (5/2, was scheduled sooner but parents had to reschedule)
Post-results Course

- PCP identified caffeine must be followed by specialist, so referred to Pulm
- Direct admitted back to us on 4/18 from Pulm given concern for respiratory compromise and need for urgent sleep study
- One episode of 101F fever 2 days prior, afebrile since
- Worsening cough
- post-tussive emesis
- No further episodes of cyanosis
- Plan per pulm: HFNC while asleep, off during day; aggressive pulm toilet; amox/clav given concern for inc risk of severe, progressive pneumonia in patients with neuromuscular disease
Final Thoughts

• rWGS did not result during admission
• No issues with process for coverage
• Still providing appropriate follow-up for anticipatory guidance from experts
• Question of now over medicalization based on knowledge?
• Associated resource costs?
What can a Pediatrician do?

Statewide PBD Case Reviews
• 3rd Thursday of each month, 12-1PM
• October 20th, 2-4 pm PBD Town Hall

Contact us:
Caleb Bupp MD - caleb.bupp@spectrumhealth.org
Andrea Scheurer-Monaghan MD - SCHEUREA@bronsonhg.org
Joseph Fakhoury MD – fakhourj@bronsonhg.org
Q&A + Resources

Project Baby Deer Website
https://www.mha.org/issues-advocacy/project-baby-deer/

Genomics 101 Resources
https://radygenomics.org/education/genomics-101/