

Baby Badger Network

MAY 14TH, 2026 12:30PM – 1:30PM



BBN ECHO: Welcome & Announcements

- Introduce yourself in the chat (name, discipline, institution)
- Please turn your camera on
- Text Attendance Code | 606-260-7097 | FOMPAG
- ECHO Session
 - Knowledge Burst – Genomic Testing in the NICU
 - Problem of Practice When language is a barrier
- Wrap Up



Knowledge Burst | Genetics Testing in the NICU

Presenter: April Hall PhD, CGC

Learning Objectives

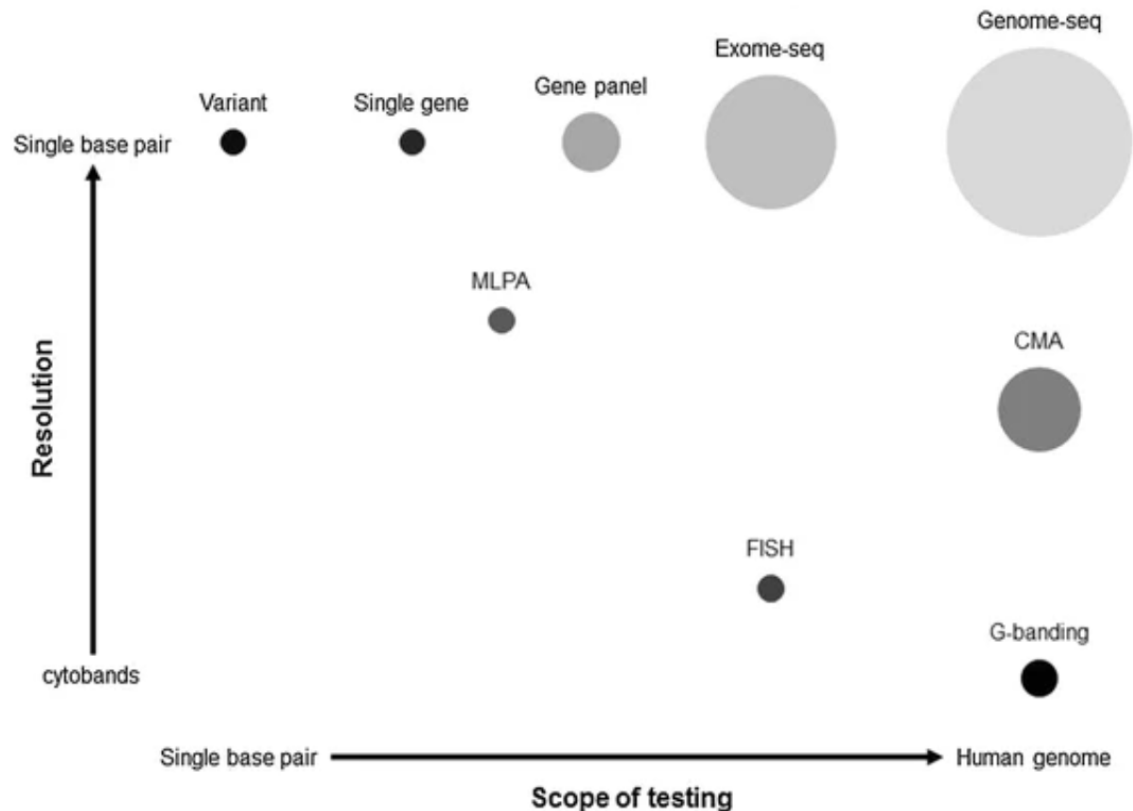
1. Compare and contrast different genomic testing options in the NICU including the type of variant each test identifies
2. Identify logistical considerations for genomic testing
3. Discuss factors influencing genomic test selection



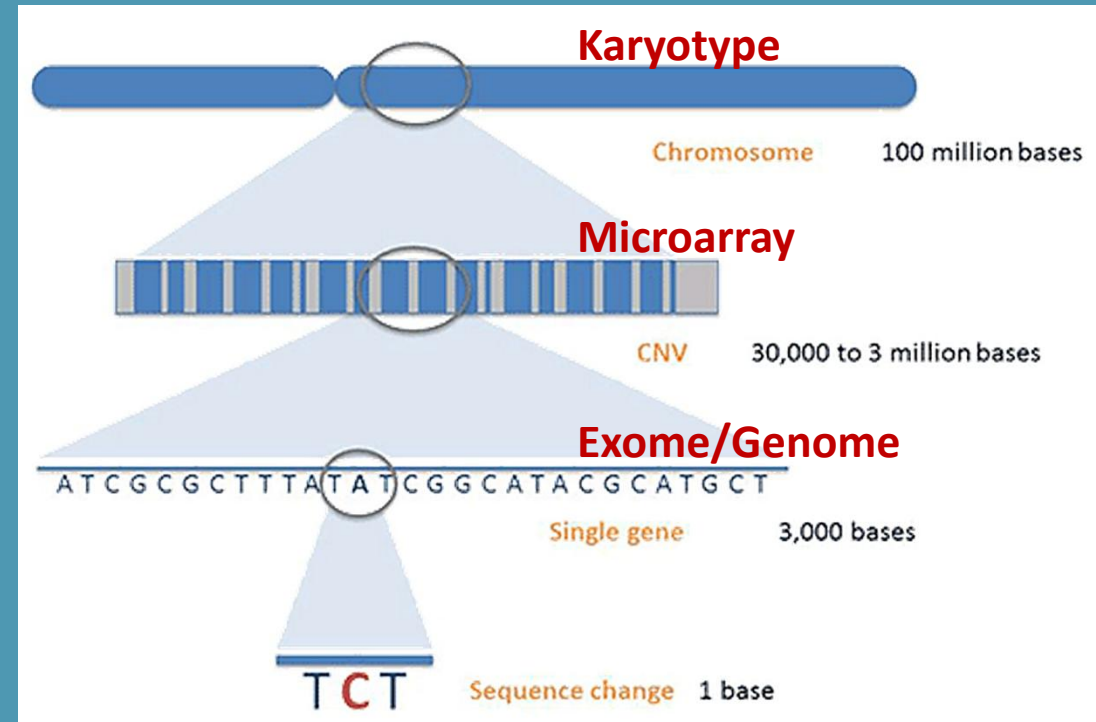
Types of Genetic/Genomic Testing

Genetic

Genomic



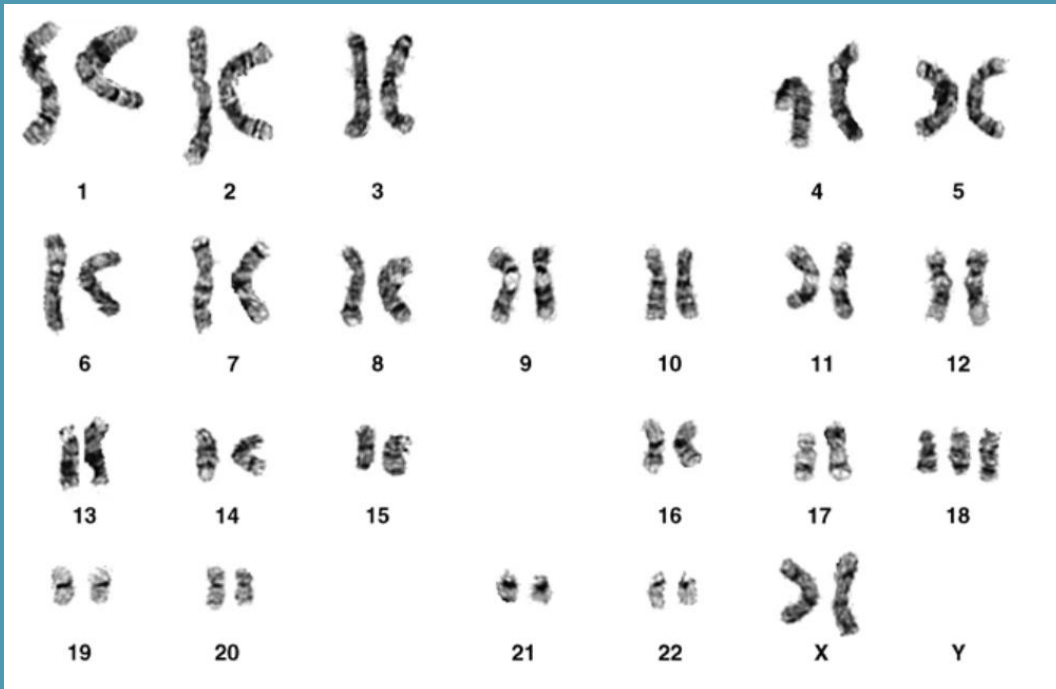
Shen & Miller, 2014



Niroshi Senaratne



Karyotype



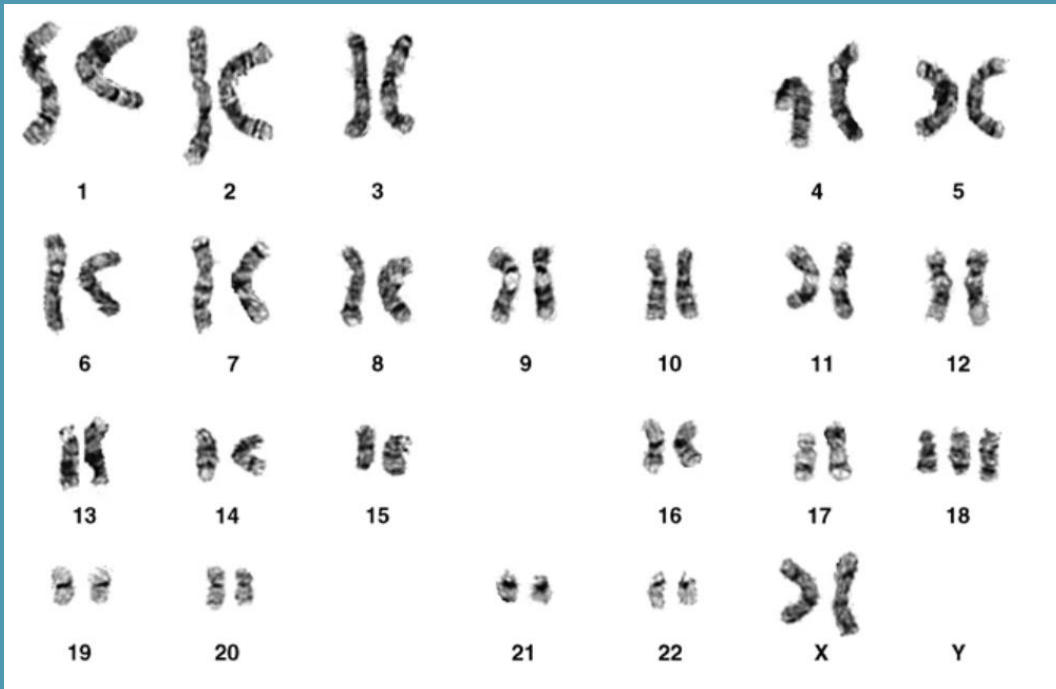
Agoston et. al, 2010

Detects:

- Aneuploidy
- Large deletions and duplications
- Chromosomal rearrangements including balanced translocations and inversions
- Triploidy



Karyotype



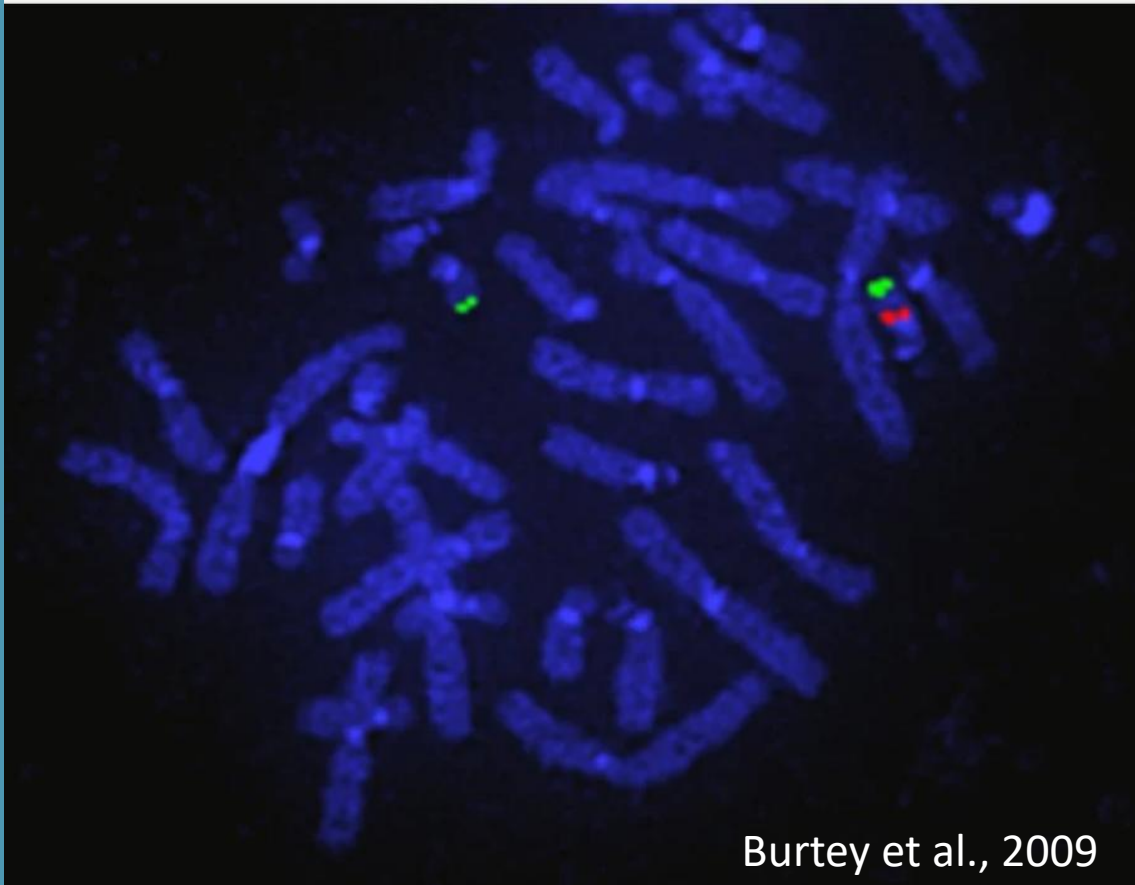
Agoston et. al, 2010

Limitations:

- Turnaround time – 1-2 weeks
- Cannot detect small changes or single gene disorders



FISH



Burtey et al., 2009

Detects:

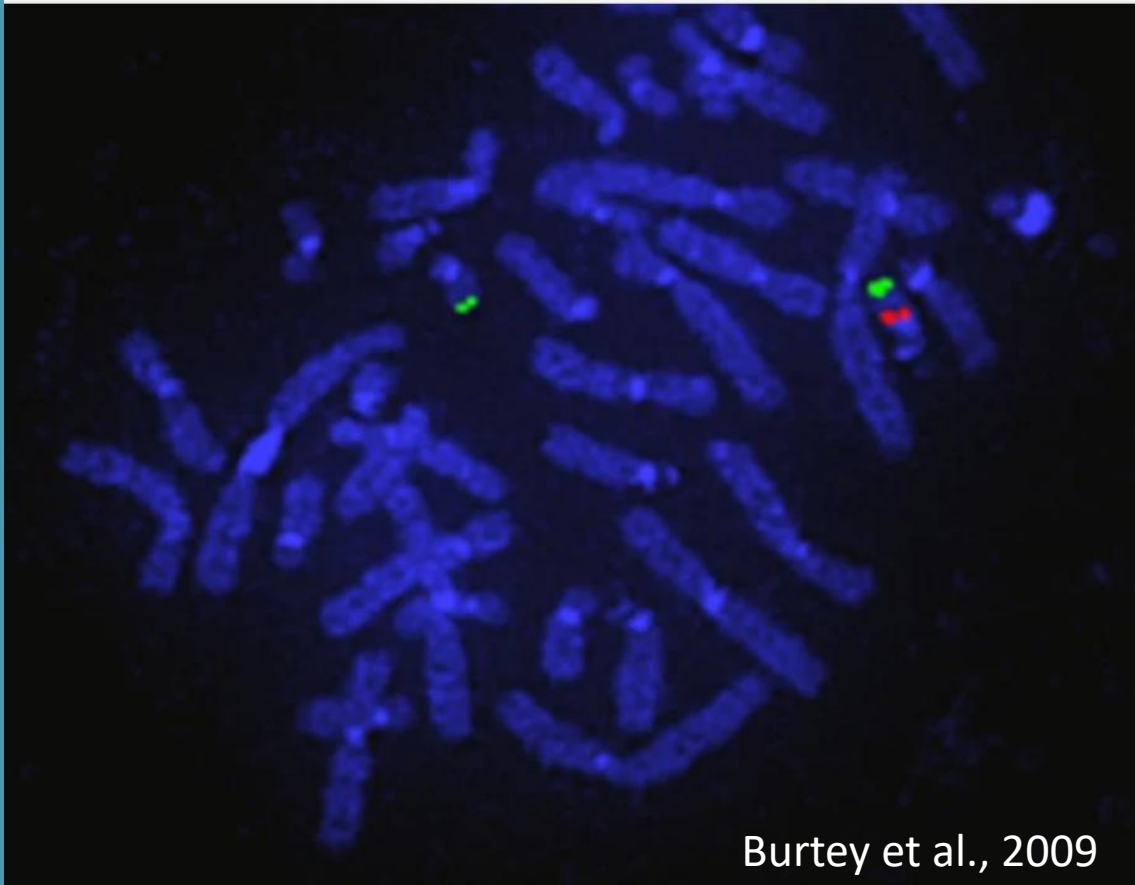
- Aneuploidy
- Microdeletions
- Low level mosaicism

Advantages

- Results in 24-48 hours



FISH



Burtey et al., 2009

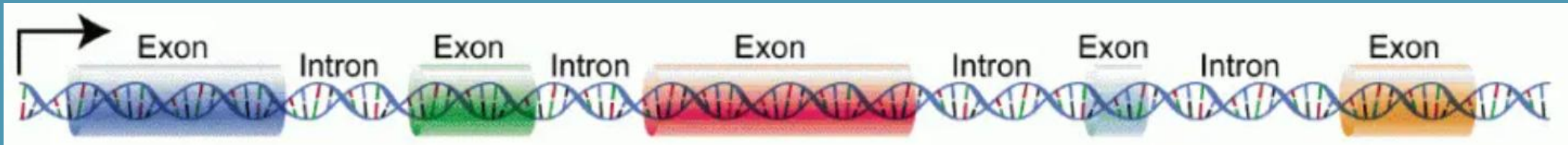
Limitations:

- All regions of the genome are not covered by probes
- Difficult to detect duplications
- Cannot detect small deletions or single nucleotide variants



Targeted Gene Sequencing

Rich Feldenberg, 2016



Can look at:

- Just exons in a gene
- An entire gene
- Multiple genes (gene panel)

Detects:

- Single nucleotide variants
- Some copy number variants

Advantages:

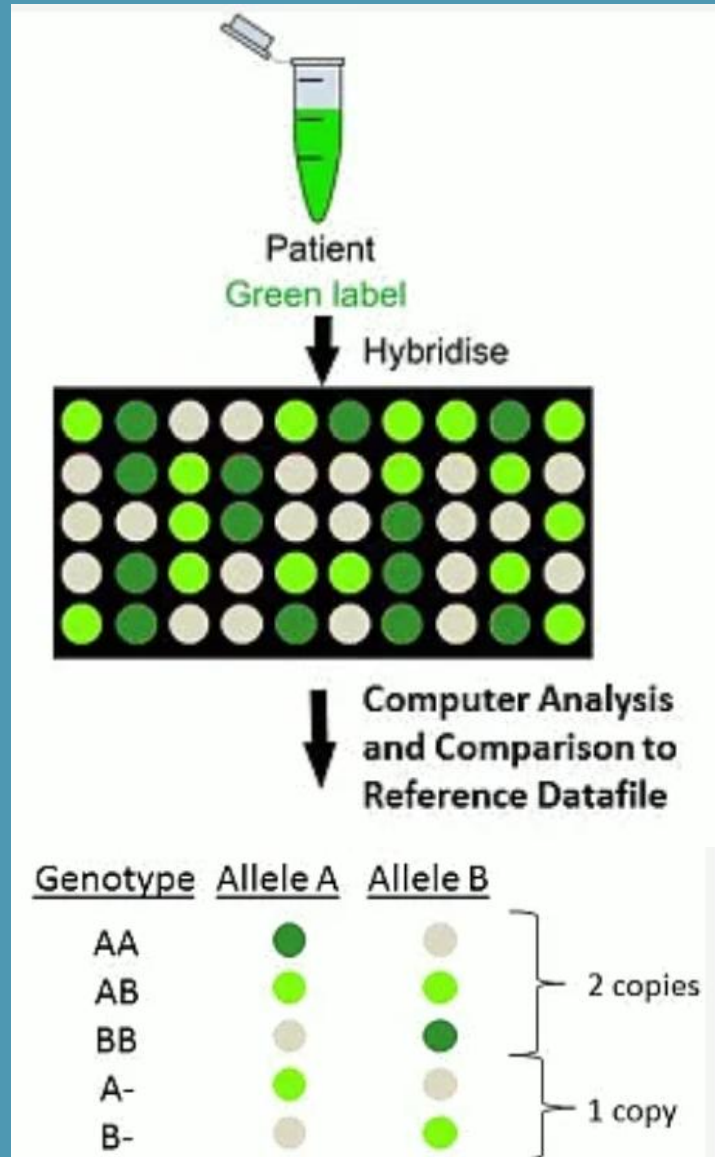
- Cost effective
- High fidelity of results

Disadvantages

- Variants of Uncertain Significance (VUS)
- Limited to one or a set of genes



Microarray



Karampetsou et. al., 2013

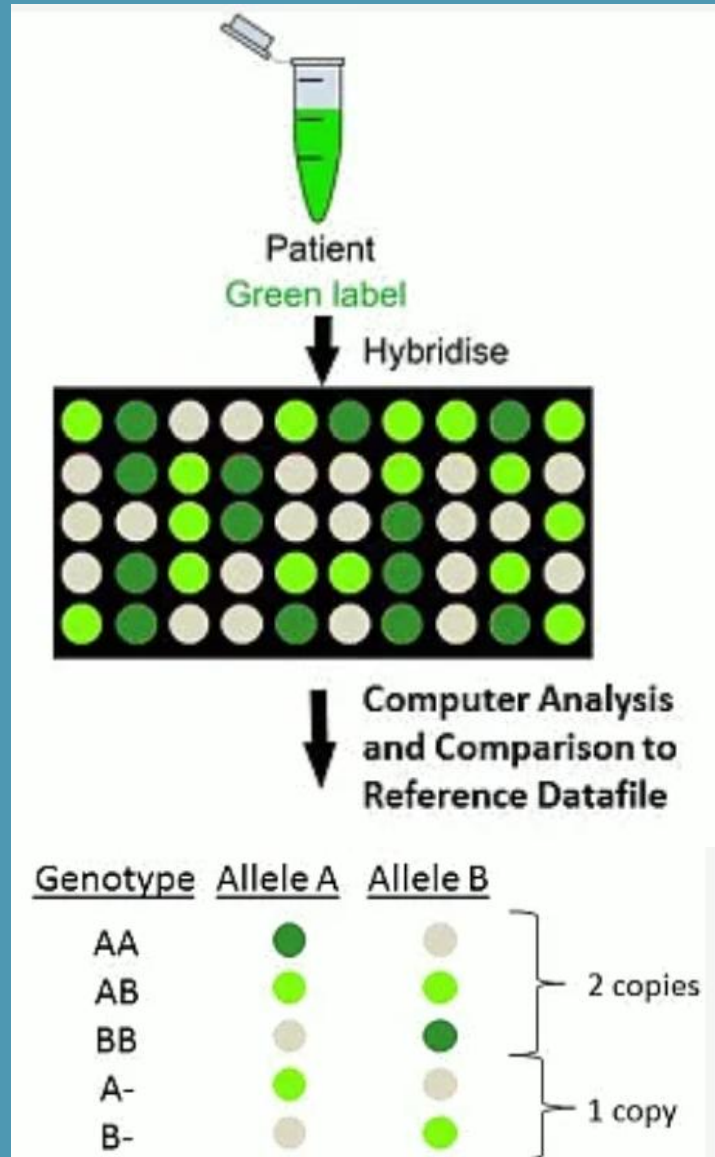
Microarray, SNP microarray, array CGH

Detects:

- Microdeletions and duplications (50-100 kb)
- Triploidy
- Consanguinity/Absence of heterozygosity
- Uniparental disomy



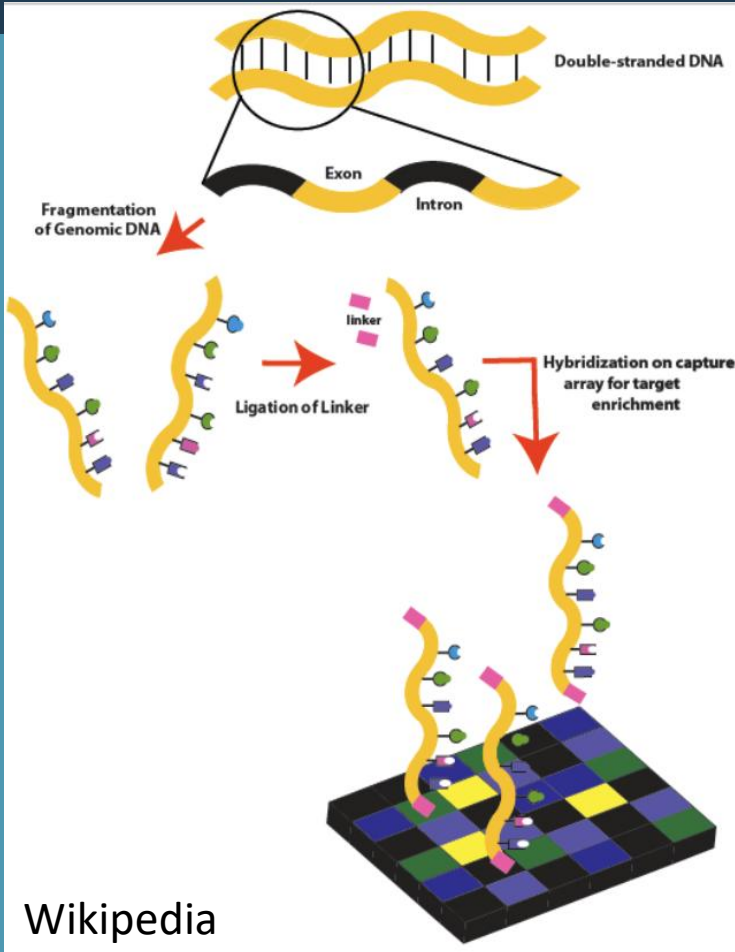
Microarray



- Limitations:
 - Copy number variant of uncertain significance
 - Incidental findings
 - Cannot detect
 - Balanced rearrangements (inversions, balanced translocations)
 - Point mutations
 - Low level mosaicism



Whole Exome Sequencing



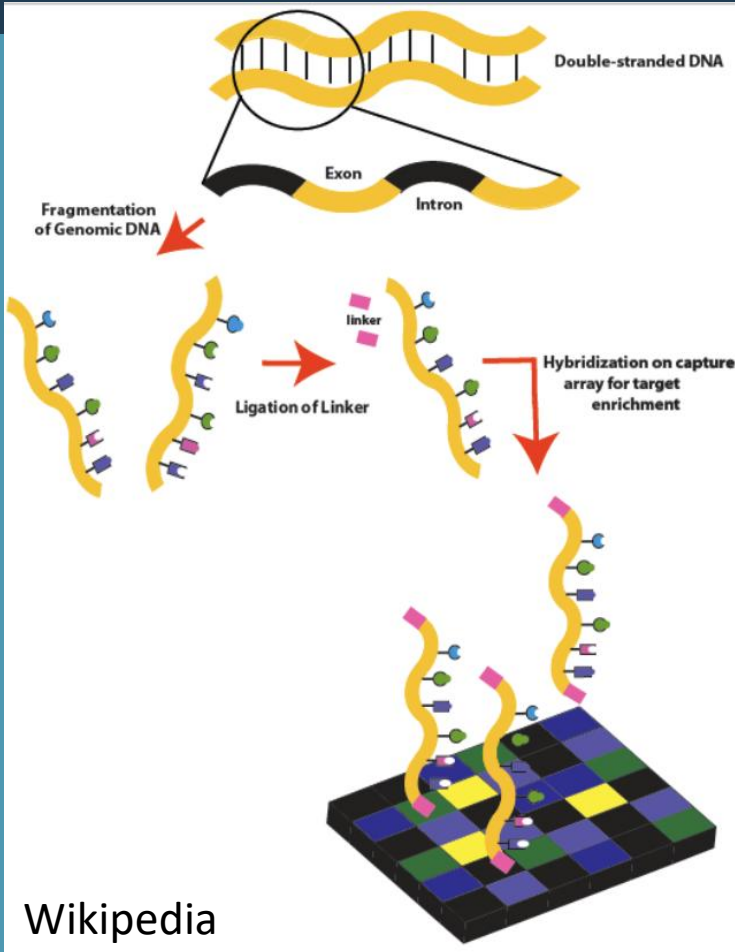
85% of disease causing mutations are in the exons

Detects:

- Single nucleotide variants
- Small insertions and deletions
- Some copy number variants



Whole Exome Sequencing

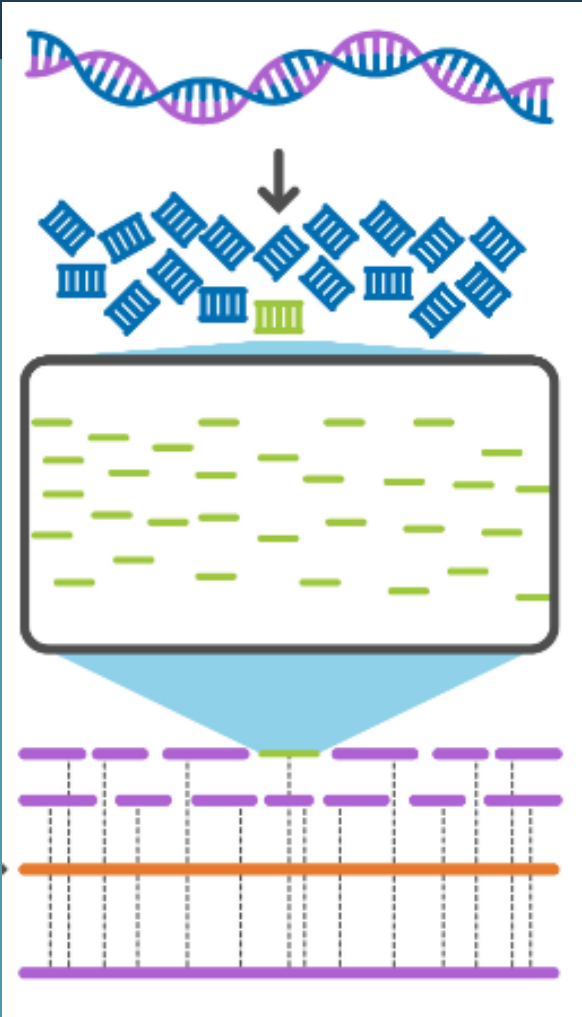


Limitations:

- Only looks at 2% of genome
- Some regions are not well covered
- Variants of uncertain significance
- Incidental findings



Whole Genome Sequencing



More comprehensive and uniform coverage

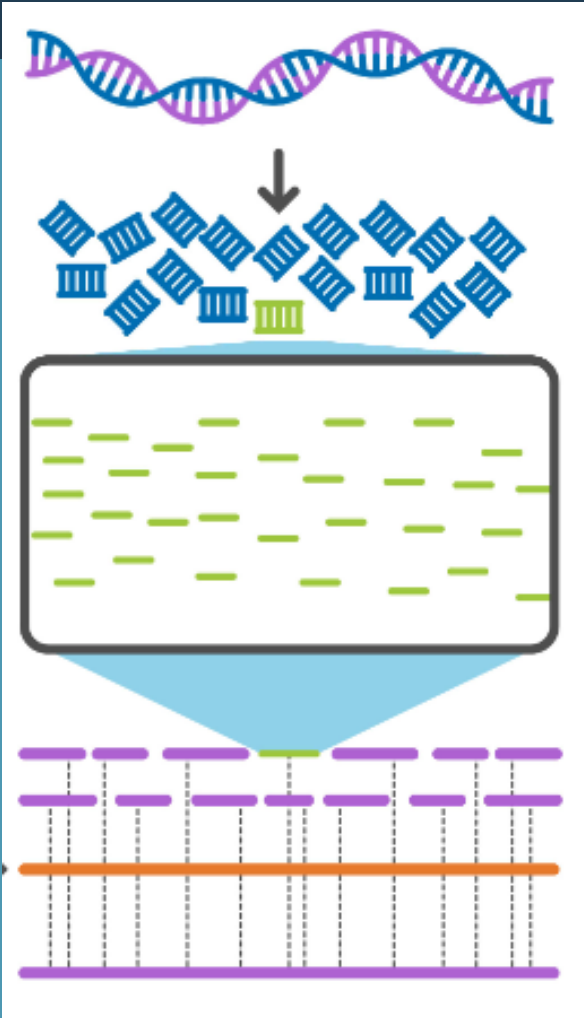
Faster turn around time

Detects:

- Single nucleotide variants
- Small insertions and deletions
- Copy number variants
- Some repeat disorders



Whole Genome Sequencing



Limitations:

- Higher number of variants of uncertain significance
- Incidental findings
- Cost



Other Genetic Tests

- Targeted Variant Testing
 - Known familial mutation
 - Specific mutation that commonly causes a particular disease
 - Mutations more common in certain ethnic groups
- Repeat disorders testing
- Methylation Studies
- Research testing
 - RNA sequencing
 - Long read genome sequencing



Problem of Practice |

- Presenter: Andrea Scheurer M.D
- Institution: Bronson Children's Hospital
- Date: May 14, 2026
- Problem/Question: Language Barriers & Genetic Testing in the NICU
 - How can we be sure families have a good understanding?
 - How do we best support families through the journey?



Case History

- 40 week male infant, 2930g
- 37 year old Mom, G3P0 (1 ectopic and 1 SAB)
- PNC from first trimester, no history of smoking, drugs, alcohol
- OB labs A pos/ID negative/Rubella Immune/GBS neg



Case History

- AMA, latent TB, Covid positive 3rd TM, GERD, hyperemesis
- Meds: PNV, Iron, Omeprazole
- US: Breech, normal growth, normal AFI, normal anatomic survey
- Normal 1 hour GTT
- Language-(Lingala)

Delivery

- Labor induced
- Delivery by C/S due to fetal intolerance of labor
- Meconium stained fluid, ruptured x 25 hours
- Nuchal cord x 1 at delivery, cord arterial gas not able to be obtained
- Baby did not require resuscitation
- Apgars 6, 8
- Initially roomed with Mom on the mother baby floor



After birth

- Newborn hospitalist team
 - Low tone
 - Unilateral undescended testicle
 - Poor PO feeding, required NG supplemental feeds
 - Mild hypoxia, required initiation of NC
 - Meconium aspiration? Covid? Sepsis?

Roomed with Mom

- Newborn hospitalist team
 - Sepsis screening labs, CBC/CRP reassuring, blood culture sent
 - SARS COV2 PCR negative
 - CXR normal
 - CUS: normal
 - CMP, Lactic acid, blood sugars, Ammonia (all normal)
 - OT consult
 - Transfer to NICU team on day 3 for congenital hypotonia, poor feeding



Transfer to NICU

- NICU admission
 - AGA male, no significant dysmorphic features, maybe lower set ears?
 - Significant hypotonia and weak sounding cry
 - Head lag and very low UE and LE tone
 - Body positioning with arms extended and laying next to his sides
 - Weak to minimal grasp
 - Slip through when held, normal suck, normal gag
 - No clonus at ankles, unable to illicit DTR at knees

What our NICU team was thinking

- Impressive exam at time of admission, but infant only 3 days old
- Clinically stable on low flow oxygen, tolerating full NG feeds
- Huge differential for congenital hypotonia
 - SMA
 - Prader Willi
 - Congenital muscular or myotonic dystrophy
 - NM junction problem
 - Others??



What our NICU team was thinking

- Communication challenges related to language barrier
- Parents took some time to see what we were seeing
- CK normal, NBS normal
- Monitored for ability to progress towards discharge
- Failed off NC for mild desaturations, echo normal
- PO ability did not improve
- Peds Neuro suggests sending rWGS



Genetic Testing Consent

bioethics

SPECIAL ISSUE: MIGRATION, HEALTH AND ETHICS |  Full Access

Language barriers and epistemic injustice in healthcare settings

Yael Peled 

First published: 09 May 2018 | <https://doi.org/10.1111/bioe.12435> | Citations: 20



CASE AND COMMENTARY
FEB 2021

Peer-Reviewed

How Should Clinicians Respond to Language Barriers That Exacerbate Health Inequity?

Jason Espinoza, MD and Sabrina Derrington, MD, MA, HEC-C

AMA J Ethics. 2021;23(2):E109-116. doi: 10.1001/amajethics.2021.109.



Consent process

- Discussion with parents (Cyracom iPOP)-days 5,6 and 11
- Parents wanted to proceed with proband only testing
- Asked for sample destruction
- Opted in to incidentals and out of research



Day of Life 13 - Collected Sample

Day of Life 17 - Result Reported

RYR1-related disorder (RYR1 pathogenic variants)

- Chromosome 19q13.2 and encodes Ryanodine Receptor 1, which releases calcium ions from the sarcoplasmic reticulum into the cytoplasm when triggered and stimulates muscle contraction

Pathogenic mutations in this gene have been associated with

- AD malignant hyperthermia
- AD and AR central core disease
- AR multi-minicore disease

CONDITION	ZYGOSITY (INHERITANCE)	VARIANT CLASSIFICATION
RYR1-RELATED DISORDERS	Heterozygous (unknown)	Pathogenic
RYR1-RELATED DISORDERS	Heterozygous (unknown)	Pathogenic
RYR1-RELATED DISORDERS	Heterozygous (unknown)	Uncertain significance



RYR1-Related Disorders

- Rare, ~ 1/10,000 people
- Range of symptoms, from almost undetectable to potentially lethal
 - Cramping, muscle pain, weakness, heat intolerance
 - Rhabdomyolysis, breathing problems, malignant hyperthermia
- Symptoms progress very slowly, or sometimes not at all
- Normal cognition
- Muscle biopsy can diagnose it but most reliable is genetic testing
- No cure
- Treatments are focused on physical/occupational therapies



Initial steps after results reported (DOL 17)

- Reviewed result with DeVos Genetics
 - Findings matched phenotype, congenital myopathy-central core
 - Self-educated OMIM/RYR-1 foundation website
 - Planned to offer telemedicine genetics consult for parents
- Reviewed result with Devos Peds Neuro
 - Formal consult planned
 - Sleep and swallow study recommended
 - Outpatient follow up with DeVos Neuromuscular clinic



Initial results discussion with parents

- Parents were frustrated with changing explanations for symptoms
- Explained no known cure, but varying phenotypes
- Parent friendly printed French clinical guideline for RYR-1 disorders
- Recommended scheduling formal genetics telemedicine consult



Resources



<https://ryr1.org>

Who We Are

The RYR-1 Foundation is a non-profit, 501(c)(3) public charity, which was started by members of the Goldberg Family, who have been affected by an RYR-1-related disease.

Currently, there is no other organization that exists solely to advocate for and serve the needs of patients with RYR-1-related diseases. The goal of The RYR-1 Foundation is to fill this much-needed void.

Who We Help

The RYR-1 Foundation works closely with individuals and families affected by RYR-1-related diseases, researchers, biotech and pharmaceutical industries, and healthcare providers from across the globe to facilitate communication in the field of RYR-1-related diseases, fund research, advance effective therapies, and optimize standards of care to enhance the quality of life in affected individuals.

Introduction

Peut-être votre médecin vous a-t-il récemment annoncé que votre enfant et/ou vous étiez atteints **d'une maladie liée au RyR-1** (maladie liée au RyR-1). Ce guide est destiné à vous aider à comprendre les divers symptômes dont vous pouvez constater la présence, ainsi que les types de soins dont vous et/ou votre famille pourriez avoir besoin. Une bonne compréhension de ces informations fera de vous un partenaire utile pour les soins qui vous seront dispensés à vous-même et/ou à votre famille.

Les mots potentiellement employés par votre médecin

CCD : myopathie congénitale à cores centraux (central core disease) - **reportez-vous au chapitre 2, « Caractéristiques cliniques des maladies liées au RyR1 ».**

CFTD : myopathie congénitale avec disproportion des types de fibres musculaires (congenital fiber type disproportion) - **Reportez-vous au chapitre 2, « Caractéristiques cliniques des maladies liées au RyR-1 ».**

CNM : myopathie centronucléaire (centronuclear myopathy) - **reportez-vous au chapitre 2, « Caractéristiques cliniques des maladies liées au RyR-1 ».**

Congénital : présent dès la naissance.

MmD : myopathie à multi-minicores (multi-minicore disease) - **reportez-vous au chapitre 2, « Caractéristiques cliniques des maladies liées au RyR-1 ».**

Myopathie : maladie musculaire dans laquelle les fibres musculaires ne fonctionnent pas normalement.

Maladies liées au RyR-1 : maladie musculaire due à une mutation du gène RyR1. Les expressions « myopathie liée au RyR-1 » et « maladie musculaire liée au RyR-1 ».

Les maladies liées à RyR-1 sont un groupe de pathologies génétiques dans lesquelles les muscles ne fonctionnent pas de manière adéquate. On utilise également le terme équivalent de « **myopathie** liée au RyR-1 ». De manière générale, les personnes atteintes d'une maladie liée au RyR-1 présentent des faiblesses musculaires ou une mauvaise tonicité musculaire. Il arrive que les enfants atteints d'une maladie liée au RyR-1 mettent plus de temps à apprendre à s'asseoir, à se déplacer à quatre-pattes et à marcher. Les personnes atteintes d'une maladie liée au RyR-1 peuvent également connaître des problèmes de moelle épinière, de muscles oculaires, de mastication, de déglutition et respiratoires. Les maladies liées au RyR-1 peuvent être à l'origine d'un large éventail de symptômes allant d'une légère faiblesse à une forme potentiellement mortelle de réaction à certaines formes d'anesthésie appelées « hyperthermie maligne » (HVM), en passant par une faiblesse grave (nécessitant parfois



Subsequent results discussion with parents

- Became clear parents had educated themselves
- Addressed several mis-understandings related to the diagnosis
- Family declined tele-medicine genetic support inpatient in favor of visit after discharge
- Parent focus became what do we do to get him home
- Began talking about recommended next steps in care plan
- Further counseling and discussions went very smoothly



Medical course after result disclosure

- Neuro consult
- PO feeds progressing some but will need tube supplementation
- Continued to require home oxygen to maintain saturation goals
- Transferred infant to level 4 NICU for inpatient sleep study
 - OSA but only mild gas exchange abnormalities
 - Pulm recommended home 0.5L/100% for home

Medical course after result disclosure

- Returned to Bronson
 - PO improved enough to support home NG, parents not interested in GT
 - Discharged home with NG/home oxygen and home pulse ox on DOL 45
 - Home nursing visits arranged
 - All follow up at DeVos: Pulm/Genetics/NM clinic/Neurodevelopment clinic

Team Reflection

- Did this family have a good understanding?
 - Ensure rapport between Neo and parents.
 - Parents agreed baby wasn't progressing and were curious as to why.
 - Introduced the idea of rWGS with follow up discussions on different days.
- How can we best support families?
 - Ask if the parents have a preferred interpreter. They had favorites!
 - Have an efficient/hospital approved work-flow for translating printed documents.
 - Show more interest in learning about their culture.
 - Greet them using their language! "Hello" = "Mbote"



Key Takeaways

rWGS workflows, consent to result are complicated at baseline

Rare diseases are challenging

Language barriers complicate communication

Establish trust with family, patience with the counseling

Be prepared to be humbled by how much we don't know

Show curiosity and respect for the family's culture



Announcements & Wrap Up

- **Text Attendance Code | 606-260-7097 | FOMPAG**
- **Claim Credit (AMA PRA Cat 1, ANCC, AAPA)**
 - Complete session evaluation (link sent via email)
- **BBN website - / XXX Add URL**
 - Knowledge Burst Recording, slides and resources
 - Problem of Practice Summary & Recommendations
- **Next Session: June 11, 2026**
 - NICU Genomic Testing Workflow Considerations

