Automation of Rapid Whole Genome Sequencing (rWGS) – The Need for Speed

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Clinical Laboratory Director, Rady Children’s Institute for Genomic Medicine
No conflict of interest

- Informed consent was obtained for this research
- Patient and parent photos, videos, and names are used with their permission
RCIGM – Who are we?

Clinical Genome Center

CLIA certification April 2017
CAP accreditation Sept. 2017

Mission: To prevent, diagnose, treat and cure childhood diseases through genomic and systems medicine research.
Rare diseases are predominantly caused by Genomic Variation and result in dire consequences for patients.

Rare disease: 1 in 2,000 affected
Ultra-rare disease: 1 in 50,000 affected

Rare diseases affect over 30 million patients in the US

- 6,517 Single gene diseases
- 22,928 Chromosomal diseases
- >1,556 Genetic diseases of unknown cause

66% of rare diseases are serious and disabling – 50% are life-limiting

75% of rare diseases affect children
95% of rare diseases lack an FDA/EMA approved treatment
450 new medicines in development for rare diseases

27% of patients with the most common 350 rare diseases will not reach their 1st birthday

Initial Focus of Pediatric Genomic Medicine

- Regional NICU, PICU and CVICU: 60% of Healthcare Cost, 5% of Children
- High Risk Children: 37% of Healthcare Cost, 45% of Children
- Healthy Children: 3% of Healthcare Cost, 50% of Children
Rapid Whole Genome Sequencing

NICU: Opportunity for Biggest Impact

Comprehensive genetic testing

Timely, targeted treatment

Better patient outcomes
The Rapid Precision Medicine Paradigm

Focus: Infants in ICUs with diseases of unknown or incorrect aetiology

### Evolution of rWGS

<table>
<thead>
<tr>
<th>Year</th>
<th>Cost</th>
<th>Time</th>
<th>Year</th>
<th>Cost</th>
<th>Time</th>
</tr>
</thead>
<tbody>
<tr>
<td>2003</td>
<td>$2.7 Billion</td>
<td>13 years</td>
<td>2009</td>
<td>$2 Million</td>
<td>1.5 years</td>
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<tr>
<td>2012</td>
<td>$13,000</td>
<td>50 hours</td>
<td>2015</td>
<td>$13,000</td>
<td>26 hours</td>
</tr>
<tr>
<td>2018</td>
<td>$8,500</td>
<td>19 hours</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

- **1st Genome**
- **1st Medical Genome**
- **1st Generation Research Prototype**
- **1st Clinical Utility Study; 2nd Generation Test**
- **1st Effectiveness Study; 3rd Generation Test**
Medical literature consistently demonstrates clinical utility

<table>
<thead>
<tr>
<th>1st Author</th>
<th>Date</th>
<th>Study Type</th>
<th>Seq Type</th>
<th>NICU and PICU Enrollment Criteria</th>
<th>Size</th>
<th>Dx Rate</th>
<th>Clinical Utility</th>
<th>Change in Outcome</th>
<th>TAT (d)</th>
<th>Savings per patient tested</th>
</tr>
</thead>
<tbody>
<tr>
<td>Saunders</td>
<td>2012</td>
<td>Cases</td>
<td>rWGS</td>
<td>NICU infants with suspected genetic disease</td>
<td>4</td>
<td>75%</td>
<td>n.d.</td>
<td>n.d.</td>
<td>2</td>
<td>$3,500</td>
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<tr>
<td>Willig</td>
<td>2015</td>
<td>Cohort</td>
<td>rWGS</td>
<td>&lt;4 mo of age; Suspected actionable genetic disease</td>
<td>35</td>
<td>57%</td>
<td>31%</td>
<td>29%</td>
<td>23</td>
<td>$1,100</td>
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<tr>
<td>Meng</td>
<td>2017</td>
<td>Cohort</td>
<td>rWES</td>
<td>&lt;100 days of life; Suspected genetic disease</td>
<td>63</td>
<td>51%</td>
<td>37%</td>
<td>19%</td>
<td>13</td>
<td>In progress</td>
</tr>
<tr>
<td>van Diemen</td>
<td>2017</td>
<td>Cohort</td>
<td>rWES</td>
<td>Infants; Suspected genetic disease</td>
<td>23</td>
<td>30%</td>
<td>22%</td>
<td>22%</td>
<td>12</td>
<td>In progress</td>
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<tr>
<td>Petrink</td>
<td>2018</td>
<td>RCT</td>
<td>rWGS</td>
<td>&lt;4 mo of age; Suspected genetic disease</td>
<td>32</td>
<td>41%</td>
<td>22%</td>
<td>n.d.</td>
<td>13</td>
<td>In progress</td>
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<tr>
<td>Farnaes</td>
<td>2018</td>
<td>Cohort</td>
<td>rWGS</td>
<td>infants; Suspected genetic disease</td>
<td>42</td>
<td>43%</td>
<td>31%</td>
<td>26%</td>
<td>23</td>
<td>$3,500</td>
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<tr>
<td>Stark</td>
<td>2018</td>
<td>Cohort</td>
<td>rWES</td>
<td>Acutely ill children with suspected genetic diseases</td>
<td>40</td>
<td>53%</td>
<td>30%</td>
<td>8%</td>
<td>16</td>
<td>$1,100</td>
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<td>Mestek-Boukhibar</td>
<td>2018</td>
<td>Cohort</td>
<td>rWGS</td>
<td>Children; PICU and Cardiovascular ICU</td>
<td>24</td>
<td>42%</td>
<td>13%</td>
<td>n.d.</td>
<td>9</td>
<td>In progress</td>
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<td>Ceyhan-Birsoy</td>
<td>2019</td>
<td>RCT</td>
<td>rWES</td>
<td>NICU neonates</td>
<td>32</td>
<td>16%</td>
<td>n.d.</td>
<td>n.d.</td>
<td>n.d.</td>
<td>In progress</td>
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<tr>
<td>Sanford</td>
<td>2019</td>
<td>Cohort</td>
<td>rWGS</td>
<td>4 months-18 years; PICU; Suspected genetic diseases</td>
<td>38</td>
<td>48%</td>
<td>39%</td>
<td>8%</td>
<td>14</td>
<td>In progress</td>
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<td>French</td>
<td>2019</td>
<td>Cohort</td>
<td>rWGS</td>
<td>Suspected genetic disease</td>
<td>195</td>
<td>21%</td>
<td>14%</td>
<td>n.d.</td>
<td>21</td>
<td>In progress</td>
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<td>Clark</td>
<td>2019</td>
<td>Cases</td>
<td>urWGS</td>
<td>Infants; Suspected genetic disease</td>
<td>7</td>
<td>43%</td>
<td>100%</td>
<td>n.d.</td>
<td>1</td>
<td>In progress</td>
</tr>
<tr>
<td>Kingsmore/Dimmock</td>
<td>2019</td>
<td>RCT</td>
<td>rWGS</td>
<td>Infants; disease of unknown etiology; within 96 hours of admission</td>
<td>94</td>
<td>19%</td>
<td>24%</td>
<td>n.d.</td>
<td>11</td>
<td>$3,000</td>
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<tr>
<td></td>
<td></td>
<td></td>
<td>rWES</td>
<td></td>
<td>95</td>
<td>20%</td>
<td>19%</td>
<td>10%</td>
<td>11</td>
<td>In progress</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>urWGS</td>
<td></td>
<td>24</td>
<td>46%</td>
<td>54%</td>
<td>n.d.</td>
<td>5</td>
<td>In progress</td>
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<tr>
<td>Baby Bear</td>
<td>2019</td>
<td>Cohort</td>
<td>rWGS</td>
<td>MediCal infants; within 1 week of admission; suspected genetic disease</td>
<td>132</td>
<td>43%</td>
<td>39%</td>
<td>In progress</td>
<td>3</td>
<td>$3,300</td>
</tr>
</tbody>
</table>

Average  | 880  | 33%     | 26%              | 14%              | $2,900            |

NICU: neonatal ICU; PICU: pediatric ICU; RCT: randomized controlled trial; rWES: Rapid exome sequencing; *£ saved per QALY
### Effect of rWGS-based precision medicine on acute healthcare utilization in six infants and three matched controls

<table>
<thead>
<tr>
<th>Subject ID</th>
<th>Presentation and modeled change in care</th>
<th>Gene</th>
<th>Time-to-diagnosis, days (method)</th>
<th>Hospital stay, Days</th>
<th>Decreased hospital stay, days (%)</th>
<th>Total cost</th>
<th>Cost avoided</th>
</tr>
</thead>
<tbody>
<tr>
<td>6011</td>
<td>Cholestasis, 1st admission for etiologic Dx</td>
<td>NPC1</td>
<td>7 (G)</td>
<td>8/15</td>
<td>15 (35%)</td>
<td>$25,278</td>
<td>$27,004</td>
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<tr>
<td>6012</td>
<td>Palliative care started DOL 250</td>
<td>ARID1B</td>
<td>26 (G)</td>
<td>250/292</td>
<td>42 (17%)</td>
<td>$1,949,438</td>
<td>$2,276,944</td>
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<tr>
<td>6014</td>
<td>Hypotonia, Avoided EMG, GA, muscle biopsy Electromyogram, GA, muscle biopsy</td>
<td>NEB1</td>
<td>7 (G)</td>
<td>45</td>
<td>2 (6%)</td>
<td>$156,914</td>
<td>$9,900</td>
</tr>
<tr>
<td>6026</td>
<td>Cholestasis and congenital heart disease</td>
<td>JAG1</td>
<td>3 (G)</td>
<td>11</td>
<td>3 (18%)</td>
<td>$50,327</td>
<td>$44,451</td>
</tr>
<tr>
<td>Control 2</td>
<td>Control Avg cost</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>$131,795</td>
<td></td>
</tr>
<tr>
<td>6041</td>
<td>Seizures. Diagnosis DOL 4</td>
<td>KCNQ2</td>
<td>4 (G)</td>
<td>18/59</td>
<td>41 (69%)</td>
<td>$79,675</td>
<td>$261,156</td>
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<tr>
<td>6053</td>
<td>Hypoglycemia. Diagnosis DOL 12</td>
<td>ABCC8</td>
<td>7 (G)</td>
<td>10/31</td>
<td>21 (68%)</td>
<td>$59,769</td>
<td>$185,283</td>
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<tr>
<td>Healthcare savings</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>$803,199</td>
<td></td>
</tr>
<tr>
<td>Cost of rWGS in 42 families</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>$674,645</td>
<td></td>
</tr>
<tr>
<td>Net healthcare savings</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>$128,554</td>
<td></td>
</tr>
</tbody>
</table>

Ongoing studies will answer the questions of how much RPM saves and how much it is generalizable

Farnaes et al 2018
Was rapid genome sequencing useful?

Physicians think RPM™ is useful

- Rapid Genome Sequencing (20% diagnosis)
- Rapid Exome Sequencing (20% diagnosis)
- Ultra-Rapid Genome Sequencing (46% diagnosis)

ClinicalTrials.gov NCT03211039
Exome sequencing: 1% of genome
Rapid: 12 days
Ultra-rapid: 2 days
So do parents

The choice to sequence did my child a lot of good:

ClinicalTrials.gov NCT03211039  28-day mortality: Rapid genome and exome sequencing 1.6%
3pm, October 24, 2017 – NICU family 243

- 8-day-old ♂ admitted from ER with Status Epilepticus
- History:
  - 23-yr G2P1 healthy mother
  - Fetal ventriculomegaly detected by ultrasound during pregnancy
  - Delivery by uncomplicated C-section 39 1/7 weeks
  - Breast-feeding well, discharged home on day of life 3
NICU family 243: Initial NICU Workup

- **Electroencephalogram:** seizures & background burst suppression
- **Brain computed tomography:** mild hypoplasia of cerebellum; Borderline lateral ventriculomegaly
- **Infection workup:** negative
- Cerebrospinal fluid lactic acid 6.3 mmol/L (normal 1.1-2.8)
- Serum creatinine kinase 1,195 U/L (normal 13-80, not in acute renal failure range)
Disease Progressed Overnight

• “Last night was rough with ongoing...multifocal seizures that continued despite...levetiracetam or phenobarbital”

  • Maximal anti-epileptic drugs
  • Worsening seizures
  • No response to phenytoin, carbamazepine
  • Midazolam drip increased until respiratory failure, emergent intubation

• “I discussed with his parents the range of outcomes I have seen with Neonatal Burst Suppression encephalopathy which usually entails limited life expectancy and at least moderate to severe developmental disabilities.”
**Diagnosis reported at 8pm October 27**

- **Disease:** Pyridoxine-Dependent Epilepsy
- **Gene:** Aldehyde dehydrogenase 7 family member A1
- **Inheritance Pattern:** Autosomal Recessive
- **Variants:** 2 pathogenic variants

<table>
<thead>
<tr>
<th>Genome variant (g.)</th>
<th>Gene variant (c.)</th>
<th>Protein variant (p.)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chr5 g.125,919,689C&gt;T</td>
<td><strong>ALDH7A1</strong> c.328C&gt;T</td>
<td>p.Arg110Ter</td>
</tr>
<tr>
<td>Chr5 g.125887751C&gt;G</td>
<td><strong>ALDH7A1</strong> c.1279G&gt;C</td>
<td>p.Glu427Gln</td>
</tr>
</tbody>
</table>

C=Cytosine; T=Thymidine; G=Guanine
Arg=Argenine; Ter=Termination Codon; Gln=glutamine; Glu=glutamic acid

_Glossary:_
- **Gene** – a sequence of nucleotides in a genome that codes for a protein
- **Recessive** – A disease expressed in offspring only when inherited from both parents
Step 11: Rapid Precision Medicine Guidance

- Supplement argenine
- Restrict dietary lysine

Following triple therapy with pyridoxine, L-arginine supplementation and dietary lysine restriction

- Electroencaphalogram normalized
- Seizures stopped

Within 36 hours

- Extubated
- All anti-epileptic drugs stopped

Discharged Home

- Meeting milestones @ 22 months of age
The Rapid Precision Medicine Paradigm Applied to Neonatal Seizures

Newborn 6 Months Before RPM

Search for etiological diagnosis

42 days empirical treatment

Unremitting seizures

Treatment Modification

Genetic Disease Diagnosis on DOL 43: Early Infantile Epileptic Encephalopathy Type 7

Natural history of disease

Profound neurologic damage

59 day hospitalization: 211,484

Rapid Precision Medicine: Carbamazepine

Minor developmental delay

17 day hospitalization: 79,675

From: Farnaes et al 2018; de novo KCNQ2 variant
Current Barriers

• Is the expense justified?
  – Is more needed?
• Genomic analysis time
  – Automated variant prioritization approaches
  – Artificial intelligence tools in development
• Limitations of genomic sequencing approaches
  – Polynucleotide repeats, regions with high homology, translocations
• Responsible consent and return of results
  – Education/outreach, collaboration with genetics services
• Decision support for molecular diagnoses
  – Education/outreach, medical fellow training
rWGS Interpretation Conundrum

Sequencing

Genomes sequenced in 17 hours

Alignment/Variant Calling

Genomes processed in 45 min

Interpretation/Reporting

Still takes hours/days
### Why is it Taking so Long? Manual Process!!!

<table>
<thead>
<tr>
<th>Steps in the Process</th>
</tr>
</thead>
<tbody>
<tr>
<td>Initial Case Tracking - test type, family members</td>
</tr>
<tr>
<td>Phenotyping - HPO terms - gene lists</td>
</tr>
<tr>
<td>Case Creation</td>
</tr>
<tr>
<td>Variant selection/filtering</td>
</tr>
<tr>
<td>Curation of variants</td>
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<tr>
<td>Confirmation ordering</td>
</tr>
<tr>
<td>LD review/reporting</td>
</tr>
<tr>
<td>Report delivery</td>
</tr>
<tr>
<td>Verbal communication</td>
</tr>
</tbody>
</table>
A lot to Interpret!

<table>
<thead>
<tr>
<th>Variant Type</th>
<th>Details</th>
</tr>
</thead>
<tbody>
<tr>
<td>Single Nucleotide Variants</td>
<td>&gt;99.8% sensitivity</td>
</tr>
<tr>
<td>Small Insertions/Deletions</td>
<td>Reported up to 40 bp</td>
</tr>
<tr>
<td>Small Copy Number Variation</td>
<td>Down to 1 kb (187 bp finding this month!)</td>
</tr>
<tr>
<td>Large Copy Number Variation</td>
<td>Microdeletion/duplication syndromes</td>
</tr>
<tr>
<td>Aneuploidy</td>
<td>Whole chromosome (trisomy)</td>
</tr>
<tr>
<td>SMN1 and SMN2 Copy Number</td>
<td>0,1,2, and &gt;3 copies</td>
</tr>
<tr>
<td>Mitochondrial Variants</td>
<td>Validated down to heteroplasmy levels of 1%</td>
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</table>

<table>
<thead>
<tr>
<th>Variant Type</th>
<th>Plans for Validation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Balanced translocations</td>
<td>Feasible for WGS data</td>
</tr>
<tr>
<td>Repeat expansions</td>
<td>Myotonic dystrophy screening</td>
</tr>
<tr>
<td>Intrinsic variants</td>
<td>Assessing current tools</td>
</tr>
<tr>
<td>Mosaic Copy Number Variants</td>
<td>Validation planning underway</td>
</tr>
<tr>
<td>Robust/automated UPD calling</td>
<td>Gathering truth samples</td>
</tr>
</tbody>
</table>
Role of the Laboratory Director in the rWGS Era
Diagnosis of genetic diseases in seriously ill children by rapid whole-genome sequencing and automated phenotyping and interpretation

Michelle M. Clark¹, Amber Hildreth¹,²,³, Sergey Batalov¹, Yan Ding¹, Shimul Chowdhury¹, Kelly Watkins¹, Katarzyna Ellsworth¹, Brandon Camp¹, Cyrielle I. Kint⁴, Calum Yacoubian⁵, Lauge Farnaes¹,², Matthew N. Bainbridge¹,⁶, Curtis Beebe⁷, Joshua J. A. Braun¹, Margaret Bray⁸, Jeanne Carroll¹,², Julie A. Cakici¹, Sara A. Caylor¹, Christina Clarke¹, Mitchell P. Creed⁹, Jennifer Friedman¹,¹⁰, Alison Frith⁵, Richard Gain⁵, Mary Gaughran¹, Shauna George⁷, Sheldon Gilmer⁷, Joseph Gleeson¹,¹⁰, Jeremy Gore¹¹, Haiying Grunenwald¹², Raymond L. Hovey¹, Marie L. Janes¹, Kejia Lin⁷, Paul D. McDonagh⁸, Kyle McBride⁷, Patrick Mulrooney¹, Shareef Nahas¹, Daehoon Oh¹, Albert Oriol⁷, Laura Puckett¹, Zia Rady¹, Martin G. Reese¹³, Julie Ryu¹,², Lisa Salz¹, Erica Sanford¹,², Lawrence Stewart⁷, Nathaly Sweeney¹,², Mari Tokita¹, Luca Van Der Kraan¹, Sarah White¹, Kristen Wigby¹,², Brett Williams⁵, Terence Wong¹, Meredith S. Wright¹, Catherine Yamada¹, Peter Schols⁴, John Reynders⁸, Kevin Hall¹², David Dimmock¹, Narayanan Veeraraghavan¹, Thomas Defay⁸, Stephen F. Kingsmore¹*
Phenome + Genome

**Human Phenotype Ontology**

- Standardized vocabulary of **phenotypic abnormalities** encountered in human disease
- Developed using the medical literature, Orphanet, DECIPHER, and OMIM
- Terms are logically defined with terms from other ontologies, e.g., GO, Uberon, and CL
- HPO currently contains approximately 12,000 terms (still growing)
Deep Phenotyping by Natural Language Processing of Epic EMR: 20 sec
Why collect a deep phenotype

- The clinical features of NICU infants do NOT correspond well with classical descriptions of their disease
- The ability to make a diagnosis is critically dependent on a full clinical description

76 children with genetic diseases; natural language processing of EHR; Text book: Mendelian Inheritance in Man
Automated provisional diagnosis:

- Nucleotides sequenced: 130,000,000,000
- Nucleotides assigned: 3,000,000,000
- Nucleotide variants: 5,000,000
- DNA changes present in <1:100 people: 750,000
- DNA changes that could cause disease: 1,000
- Provisional diagnosis: 1

*Genetic diseases assoc. with seizures: x 1,250*
Next Steps and Where Does AI/NLP Come in?

<table>
<thead>
<tr>
<th>Steps in the Process</th>
<th>Automation/NLP</th>
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<td>Initial Case Tracking - test type, family members</td>
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<td>AUTOMATION</td>
</tr>
<tr>
<td>Verbal communication</td>
<td>NLP/AI – RPM GUIDANCE</td>
</tr>
</tbody>
</table>
Current network and key collaborators

Coalition of 600 rare disease organizations

1200 NICUs in 30 countries working to continuously improve neonatal care
Early, comprehensive diagnosis with therapy guidance will increase orphan drug markets

**Ultra-rapid genome sequencing**

**Rapid genome sequencing**

**Standard genetic tests**

urWGS ordered on the day of admission with 1-2 day time to result is optimal
We are developing a healthcare delivery system for national implementation of Rapid Precision Medicine

1. Ultra-Rapid Diagnostic Evaluation
   - 1 day to result
   - Semi-automated interpretation
   - 3,000 cases / year
   - State-of-the-art diagnostic performance

2. Stakeholder, Health System, and Patient/Family Engagement
   - Engages all stakeholders
   - Genomic consult service
   - Context-specific implementation
   - Simplified, non-expert ordering
   - Automated deep phenotype extraction

3. Precision Medicine Delivery and Follow-up
   - Results effectively communicated to non-expert ICU teams & parents
   - Management guidance to change Rx before discharge
   - Implications understood by parents
   - Precision medicine follow-up clinic

4. Rare disease therapeutic innovation by analysis of longitudinal knowledgebase of 10,000 cases

Virtuous circle: Implementation identifies unmet needs that drive innovation
Acknowledgements: *A Deo lumen, ab amicis auxilium*

**Executive Team**
Stephen Kingsmore MD, DSc
Wendy Benson
Charlotte Hobbs, MD, PhD
David Dimmock MD

**Leadership**
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Lauge Farnaes MD, PhD
Karen Garman EdD, MAPP
Shareef Nahas PhD
Julie Reinke
Grace Sevilla, APR
Mari Tokita MD
Ray Veeraraghavan PhD
Russell Nofsinger, PhD

**Illumina**
Kevin Hall
Haiying Grunenwald

**Clinical Genome Center**
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Terence Wong PhD
Meredith Wright PhD

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Lisa Salz MS, LCGC
Kelly Watkins MS, LCGC

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Jeanne Carroll MD
Tina Chambers PhD
Michele Feddock, CCRP
Jennifer Friedman MD
Joseph Gleeson MD, PhD
Iris Reyes
Jonathan Sebat PhD
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