Claritas Genomics: Transforming a Hospital-based Molecular Lab into a Next-Generation Pediatric Diagnostic Company

Patrice M. Milos, PhD
President, Chief Executive Officer
Themes Emerging This Week

“Innovate, Transform and Partner”
Our Mission

To be the world’s leading provider of the next-generation of pediatric diagnostics
Our Mission

• Commitment to providing the highest quality and most clinically useful genetic information to guide pediatric care

• Increase scale to allow broader access to high quality information, including expertise from the pediatric hospitals
Why Claritas Genomics?
Pediatrics First - Our Knowledge of Disease Genetics

![Graph showing the number of variants reported on HGMD from 1975 to 2015](image)

**Figure 1.** Total human disease variants reported on Human Gene Mutation Database (HGMD) according to the year of publication.

**Lentiviral Hematopoietic Stem Cell Gene Therapy in Patients with Wiskott-Aldrich Syndrome**

Alessandro Aiuti,1*2,4,6 Luca Biasco,1* Samantha Scaramuzza,7 Francesca Ferrua,2,3,5 Maria Pia Cifolese,5 Cristina Baricordi,1 Francesca Dionisi,1 Andrea Calabria,1 Stefania Giannelli,1 Maria Carmina Castello,1,3 Marita Bosticardo,1 Costanza Evangelio,2,5 Andrea Assandri,2,5 Miriam Castraghi,2 Sara Di Nunzio,1 Luciano Callegaro,1 Claudia Benati,1 Paolo Rizzardi,1 Danilo Pellin,1 Clelia Di Serio,1 Manfred Schmidt,1 Christof Von Kalle,1 Jason Gardner,1 Nalini Mehta,1 Victor Nedeva,1 David J. Dow,1 Anne Galy,5 Roberto Miniero,1 Andrea Pinoci,1 Ayse Metin,1 Pinaki Banerjee,5 Jordan Orange,5 Stefania Galimberti,5 Maria Grazia Valsecchi,5 Alessandro Biffo,1,2,5 Eugenio Montini,1 Anna Villa,1* Fabio Ciciri,2,5 Maria Grazia Roncarolo,1,2,4,6 Luigi Naldini,4,6

*Canadian Journal of Cardiology (2013) 29:934-939
Molecular Diagnostics Based on Genomics is Growing

**Market Growth**
- $5 billion: 2010 U.S. spending on molecular diagnostics*
- 12 - 15% per year annual growth*
- $15 - $25 billion: overall in US by 2021*

**The Essence of Medical Genomics**
- Integrate genome and clinical information
- Stratify patients and select therapies
- Long-term relationship between patient and health care system
- Enable more precise care

**Changing Landscape**
- Cost of genome sequencing: $100M in 2001 now $~2000
- Clinical use now feasible

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<thead>
<tr>
<th>Sample</th>
<th>→</th>
<th>Processing</th>
<th>→</th>
<th>Analysis</th>
<th>→</th>
<th>Reporting</th>
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* CLARITAS GENOMICS
Medical Genomics is a Pediatric Hospital Imperative

Hospitals need:
• Excellent and cost effective molecular testing
• Specimens and data for research
• Patients with heritable disorders
• Collaborations with other institutions
• Funding to support rapid growth

However, large scale investments to build infrastructure needed to meet goals.

**Boston Children’s Hospital’s bold decision:**
Spun out the DNA Diagnostic Lab as commercial entity
• Synergy between the clinical lab and research goals
• Allows additional investment
• Strategic partnerships can reduce costs
• Access volume from patients from outside BCH
• Create economies of scale
Claritas Genomics is Born – February 2013

*The next-generation of pediatric diagnostics*

Commercial reference laboratory performing genetic testing services

- Best quality services, from pre-ordering to post-report
- Testing for any condition
- Single gene tests to microarrays to exomes to genomes
- All technology platforms
- Based on the expertise at pediatric hospitals
# Executive and Board Leadership

## Executive Team

<table>
<thead>
<tr>
<th>Name</th>
<th>Current or Past Roles</th>
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<tbody>
<tr>
<td><strong>Patrice Milos, Ph.D.</strong></td>
<td>• Former SVP, CSO in Single Molecule Sequencing technology and applications</td>
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<tr>
<td><strong>President, CEO</strong></td>
<td>• Executive Director at Pfizer Global R&amp;D; Industry leader in pharmacogenomics</td>
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<tr>
<td></td>
<td>• Established track record in securing partnerships and funding</td>
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<tr>
<td><strong>Mary Ellen Cortizas, JD</strong></td>
<td>• Co-founder, Claritas Genomics</td>
</tr>
<tr>
<td><strong>Chief Operating Officer</strong></td>
<td>• Administrative Director of Laboratory Medicine, Boston Children’s Hospital</td>
</tr>
<tr>
<td><strong>Nurjana Bachman, PhD</strong></td>
<td>• Co-founder, Claritas Genomics</td>
</tr>
<tr>
<td><strong>Chief Business Officer</strong></td>
<td>• Business Development and Strategic Alliances, Boston Children’s Hospital</td>
</tr>
<tr>
<td><strong>Todd Krueger</strong></td>
<td>• Head of BD, Molecular Medicine, Life Technologies</td>
</tr>
<tr>
<td><strong>Chief Financial Officer</strong></td>
<td>• Co-Founder, CFO, Fluidigm Corporation</td>
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## Board of Directors

<table>
<thead>
<tr>
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<th>Current or Past Roles</th>
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<tbody>
<tr>
<td><strong>Bob Higgins (Chairman)</strong></td>
<td>• Founder Highland Capital Partners, Faculty, Harvard Business School</td>
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<tr>
<td><strong>David Margulies, MD</strong></td>
<td>• Executive Director, Gene Partnership Program Boston Children’s Hospital</td>
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<tr>
<td></td>
<td>• Founder, CEO, Correlagen Diagnostics</td>
</tr>
<tr>
<td></td>
<td>• Co-founder Claritas Genomics</td>
</tr>
<tr>
<td><strong>Ron Andrews</strong></td>
<td>• President, Medical Sciences, ThermoFisher</td>
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<td></td>
<td>• CEO – Clarient</td>
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<tr>
<td></td>
<td>• President, Roche Molecular</td>
</tr>
<tr>
<td><strong>Stuart Novick</strong></td>
<td>• Legal Counsel, Boston Children’s Hospital</td>
</tr>
<tr>
<td><strong>Clay Patterson</strong></td>
<td>• VP, Managing Director, Cerner Capital</td>
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Our Investors – Foundation for Success

- **Boston Children’s Hospital**
  - Founding institution
  - World-leading pediatric hospital

- **Cincinnati Children’s Hospital**
  - Founding network partner
  - World-leading pediatric hospital

- **ThermoFisher**
  - Leading provider of DNA sequencing technology

- **Cerner**
  - World leader in healthcare information technology

Unique skill sets, commitment and infrastructure combine to bring medical genomics to the clinic broadly and improve health for all children.
## A Comprehensive Approach to Meet Our Customers’ Needs

<table>
<thead>
<tr>
<th>Partner</th>
<th>Need</th>
<th>Claritas Service</th>
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<tbody>
<tr>
<td><strong>Patients</strong></td>
<td>Fast, reliable answer</td>
<td>Genetic testing with consistent interpretation</td>
</tr>
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<td><strong>Clinicians</strong></td>
<td>Support, streamlined process</td>
<td>Pairing test with clinical need, easy ordering and billing</td>
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<tr>
<td><strong>Hospitals</strong></td>
<td>One stop shopping, best value, integrated reporting</td>
<td>Advice navigating testing landscape and consolidating send-out</td>
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<tr>
<td><strong>Payers</strong></td>
<td>Utility, value</td>
<td>Right test, strong clinical justification</td>
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<tr>
<td><strong>Researchers and health systems</strong></td>
<td>Large numbers of well-characterized samples, scalability to enable discovery</td>
<td>Research, data network</td>
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TESTING SERVICES

Fast, Reliable Answers

- Test menu – over 110 tests currently offered
  - Single gene - Now
  - NGS Panel for DMD – Available Now
  - Proprietary CNV Array – Optimized for Autism
  - Proteinuria Panel – Summer 2014
  - NGS exome-based tests – Summer 2014

- Assays arising from the clinic, developed for clinicians

- CLIA facility, using all technology platforms

- Goals: Affordability, efficiency, analytic automation, scalability
Navigating the Menu

Test selection is tied to clinical presentation:

- Takes away some of the guess work
- Upfront consultation to ensure that correct test is ordered
- Support justifying prior authorization

Example – test selection transitioning...

...from this:

- 1p36 deletion syndrome
- 1q21.1 deletion/duplication
- 15q13.2-q13.3 deletion
- 16p11.2 deletion/duplication
- 17q12 deletion
- 17q21 microdeletion syndrome
- Achondroplasia
- Acute megakaryoblastic leukemia
- Alagille syndrome
- Angelman syndrome
- etc...

...to this:

Check all that apply:
- Failure to thrive
- Muscle weakness
- etc...
Claritas Differentiator: Building a Network

- Connecting the best pediatric hospitals to share expertise
- Individual instances are rare, but across a network become common
- Overcapacity in the pediatric labs leads to inefficiency in testing.
- Sharing infrastructure will allow specialization and scale.

*Allow Pediatric Hospitals to Focus on Clinical Care*
Claritas Model

The US Pediatric Hospitals and their Molecular Diagnostic Labs
Equity and Infrastructure Sharing

- Cincinnati Children’s
- Boston
- West
- South
- Southeast
- Mid-Atlantic

Commercial subsidiary
Molecular testing lab
A Unified Approach to Product Development

Genetic testing is currently fragmented

Dysmorphology, Developmental delay
Neurology
Hematology
Metabolic
Cardio
Skeletal
Pharm
Pulm
ENT
Building Regions Of Interest to Deliver Clinical Exome Defined Subpanels

Clinical Exome (25,000 Genes) → Neurome (614 genes) → Inherited Disease Genes (3000 genes)

Strategy: Deep Insight into Clinical Validity
Phenotype: Focus on Regions of Importance
Experts: Interpret and Report

Measure
Report
2-3Q2014
AmpliSeq Research Exome

RESEARCH PRODUCT
• Claritas is a Certified Service Provider of the Ion Torrent AmpliSeq Research Exome
• Turn around time 4-6 weeks
• Various data formats available for data return
• $800 per specimen, volume discounts available
The Path to a Clinical Exome
Deriving Benefit from the VA Program

Initiate ROI
Epilepsy + Seizure + NMD

Exome Based ROI “Neurome”

Combine Variants
Dual NGS Platforms

Methods
Automate

VA Research Exome

CONFIDENTIAL
Current Lab Workflow

Order
Specific question

Lab Processing
measurement

Result
number
Claritas Future Workflow

Complex clinical information → Order → Lab Processing → Reporting → Complex report

- Analytics
- Sequencer
- Device Integration

Cerner
Expert Network

Additional customers
• Hospitals without a lab
• Adult hospitals
• Non-specialists

• Claritas as a platform to facilitate access to expertise at the pediatric hospitals
• Helps patients find local experts
• Drives volume to the hospitals
Pediatric Proteinuria
Validating Our Expert Network Approach

• **Expert**: Collaborative Development with pediatric hospital chief of Nephrology with expertise in disease management, interpretation of disease variants and with global sample repository with well-characterized genetic/phenotype correlations

• **Clinical Purpose**: Focused panel based on specific phenotypic presentations in pediatric nephrotic syndrome, in this case proteinuria; treatment decisions based on findings in large % of patients

• **Design**: 29 genes, 3 non-coding regions, PGM NGS, design supports Sanger confirmation, Automation

• **Software**: Infrastructure build supporting workflow
Pediatric Research Network

- Research network
- Data sharing
- Sample sharing
- Cohort selection
- i2b2 and tranSMART as infrastructure
Send-out Management

Additional customers
• Hospitals without a lab
• Adult hospitals
• Non-specialists

Claritas
• Be single point of contact for ordering, specimen processing, result return
• Apply best practices around utilization
• Negotiate volume discounts
• Connect IT systems
• Build use cases for payers

Reference labs
• Commercial
• e.g. LabCorp, Prevention, GeneDX, etc.
• Academic
The Send-Out Product
A Fundamental Value for Pediatric Hospitals

Claritas Manages Institutions’ Molecular Testing Send-out

- Partner Institutions will send all of their molecular send-outs to Claritas
  - Samples taken in as blood, Claritas extracts DNA
  - No additional cost for service
- Claritas performs test
  - In-house
  - Ships a portion of the DNA to Reference Labs
- Claritas negotiates volume discounts at 3rd party labs
- Claritas and partner experts decide on the “best” lab for each test
  - Optimal utilization of services
  - Quality, turn around time, price, etc
A Unified Approach to Working with Hospitals

**Equity Partner: Cash Investment/Contributing the Laboratory:**
- Share infrastructure, specialize and benefit from economies of scale

**Expert Network:**
- Provide clinical interpretation and connection to medical care

**Genomic Research Network:**
- Data sharing, allows quicker aggregation of results for discovery and clinical trial services
- Institution owns data and has first right to publish
- Learn and share best practices

**Reference Lab Network:**
- Send-out partner (one-way and reciprocal), consolidate send-out and negotiate volume discounts with partner labs

**Customer:**
- Order genetic testing services, run assays for clinical trials
Where We Are Today

- Spun out the lab and raised Series A round.
- Partnered with Cincinnati Children’s (ongoing conversations with 15+ other pediatric hospitals)
- Partnered with ThermoFisher and Cerner Corporation to provide end-to-end solution
- Awarded $9M Veteran’s Administration contract for 18,000 exomes to build scale/technology
- Launching clinical next-generation sequencing tests that unify the fragmented market
- Building Big Data infrastructure (compute, analysis, automation)
Benefits for the Pediatric Hospitals

• Focus on clinical outcomes and advancing research, not duplicating infrastructure that can be shared
• Stay current with technological advance with minimal investment
• Claritas handles commercialization, marketing and sales to drive volume
• Share data, informatics tools
• Shareholder in Claritas and benefit from revenue growth
• Platform to distribute expertise and assays
• Collaboration with other organizations to address the challenges in pediatric genetic diagnostics
Find us at www.claritasgenomics.com

Who we are

Claritas is a genetic diagnostic laboratory that has the goal of providing the highest quality testing services for diagnosis of pediatric disorders. Originating as the in-house genetic testing lab at Boston Children’s Hospital, Claritas was launched as a stand-alone entity in February 2013, and in October 2013, Cincinnati Children's Hospital joined us, beginning the formation of a genomic network of pediatric hospitals. Claritas aims to support institutions as they integrate genomics into the practice of medicine.

Testing Services

Claritas offers services that support all aspects of the clinical testing experience, from test ordering to providing the most
Claritas Genomics is committed to helping you

Ask the right questions
Choose the right test
Get the right answers
Find the right support