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Ultragenyx has not influenced the development of the content for our presentation.

Continuing Education Credits are not offered for this presentation.
INTRODUCTION:
Kristin Voorhees, MA
Senior Manager,
Patient Advocacy Ultragenyx
Our Commitment and Focus on Rare and Ultra-Rare Diseases

Kristin Voorhees, MA
Patient Advocacy
Global Medical Affairs
April 4, 2019
Ultradeutyx Pharmaceutical

- Ultradeutyx is a biopharmaceutical company committed to bringing to patients novel products for the treatment of rare and ultra-rare diseases, with a focus on serious, debilitating genetic diseases.

  Headquartered in Novato, California

  Other offices:
  - CA Bay Area
  - Massachusetts
  - Europe
  - Latin America

- 500+ employees
- Publicly-traded

TRANSFORMING GOOD SCIENCE INTO GREAT MEDICINE FOR RARE GENETIC DISEASES.
Our Commitment and Focus
Our History and Commitment

- Founded by Emil Kakkis, MD, PhD in 2010

- Focused on addressing diseases with high unmet medical need and clear biology, for which there are no approved therapies
  - Currently studying 14+ rare and ultra-rare diseases indications, including inborn errors of metabolism

- Accelerating diagnosis is critical, which often requires early detection among newborns; for example:
  - Fatty acid oxidation disorders (FAODs) are listed on the Recommended Uniform Screening Panel (RUSP) for newborn screening
  - Non immune hydrops fetalis (NIHF) can be one of the earliest symptoms of Mucopolysaccharidosis VII (MPS VII, Sly syndrome), with about 40% of patients who have presented with the condition\(^1\)

- Access to investigational therapies
  - Ultragenyx provides access to investigational therapies through its early access program. This global program evaluates patient needs and provides investigational therapies to qualified patients around the world, based on individual circumstances and clinical need.
Rare Disease Facts

30 Million Americans

350 Million Worldwide

The Impact
Rare diseases impact more people than Cancer & AIDS combined.

No FDA Approved Cures
Only 5% of rare diseases have an FDA approved drug treatment.

What is Considered “Rare”?
In the United States, a condition is considered “rare” if it affects fewer than

www.globalgenes.org

TRANSFORMING GOOD SCIENCE INTO GREAT MEDICINE FOR RARE GENETIC DISEASES.
**Rare Disease Facts**

**RARE DISEASE TYPES**
- Identified rare diseases, with more being discovered every day.
- 7,000+

**THE CAUSE**
- 80% of rare diseases are caused by faulty genes.

**RARE DISEASE EFFECT**
- 30 million Americans
- If all of the people with rare diseases lived in one country, it would be the world's 3rd most populous country.

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**World RARE Disease Day**

The first World Rare Disease Day was organized and held on February 28, 2008 by European organization, EURORDIS and its Council of National Alliances. World Rare Disease Day was created because treatment for many rare diseases is insufficient, as are the social networks to support individuals with rare diseases and their families. On World Rare Disease Day and leading up to this day, people around the world come together to raise awareness of rare diseases and the impact on those affected.

www.globalgenes.org

- Infographics provided by Global Genes

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TRANSFORMING GOOD SCIENCE INTO GREAT MEDICINE FOR RARE GENETIC DISEASES.
Visit Our Table to Learn About…

- **Role of Patient Advocacy at Ultragenyx**
  - Develop and share education, resources and information for patients and families
  - Partner with patient organizations to support patients and caregivers
  - Facilitate connections to improve the lives of everyone affected by rare diseases

- **Ultragenyx’s disease areas of focus, as well as how we help to raise awareness of the importance of early detection of rare diseases among newborns**

- **Where to get more information about rare diseases**
  - Global Genes: [www.globalgenes.org](http://www.globalgenes.org)
  - National Organization for Rare Disorders (NORD): [www.rarediseases.org](http://www.rarediseases.org)
  - Rare Disease Day: [www.rarediseaseday.org](http://www.rarediseaseday.org)
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<thead>
<tr>
<th>Name</th>
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<tr>
<td>Julie Howard</td>
<td>Parent Advocate, Mother to Spencer</td>
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<td>Mary Tucker</td>
<td>Executive Secretary to Connecticut Newborn Screening Committee, The Div, of Medical Genetics UConn</td>
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<tr>
<td>Jacquelyn Halliday</td>
<td>Senior Genetic Counselor, Women and Infants Hospital of Rhode Island</td>
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4 AREAS

- Screening
- Assessment
- Treatment Protocols
- Medical and Developmental Services

TRANSFORMING GOOD SCIENCE INTO GREAT MEDICINE FOR RARE GENETIC DISEASES.
SCREENING

- Antenatal
- Newborn Screening
- Time of Diagnosis
- Informing Families
ANTENATAL
NEWBORN SCREENING
TIME OF DIAGNOSIS
INFORMING FAMILIES

TRANSFORMING GOOD SCIENCE INTO GREAT MEDICINE FOR RARE GENETIC DISEASES.
ASSESSMENT
INFORMING
FAMILIES
TREATMENT PROTOCOLS
MEDICAL and DEVELOPMENTAL SERVICES
GENETIC COUNSELING

What is a genetic counselor?
As the study of genetics grew over the years, it became clear there was a need for professionals who understood genetics and could guide and support patients as they made decisions about genetic testing and their healthcare.

In 1971, the profession of genetic counseling was born as the first class graduated from Sarah Lawrence College.
GENETIC COUNSELING

What is a genetic counselor's training?
Bachelors degree in biological science, a social science or related field.

A master’s degree from an accredited program.

Certified through the American Board of Genetic Counseling (ABGC).

22 states have licensure.
Who is a genetic counselor?

- Specialized healthcare professionals
- Experts in genetics AND communication
- Patient advocates
- Educators
- Researchers
- Administrators
Where will you find genetic counselors?
ROLES AND SPECIALITIES

TRANSFORMING GOOD SCIENCE INTO GREAT MEDICINE FOR RARE GENETIC DISEASES.
PRENATAL GENETIC COUNSELING

I work with families who are pregnant or planning a pregnancy.

We counsel individuals regarding available prenatal screening and diagnostic options.

When a fetal disease or malformation is detected or suspected we help the family understand the diagnosis, likely prognosis, further testing options, and facilitate appropriate pediatric consultations.
PRENATAL GENETIC COUNSELING

We counsel families afflicted with a known genetic condition who are considering preimplantation genetic testing (PGT) either for a chromosome abnormality or singe gene disorder.

Becoming much more common as more gene mutations are identified through testing of affected family members.
Barriers to obtaining genetic counseling services
REMOVING BARRIERS

- Increasing the genetic counseling work force
- Continued efforts for reimbursement for services
- Increased education to non-genetic providers
- Telegenetics to increase access for patients and families
- Research regarding existing barriers to services and access disparities
BARRIERS - PROVIDERS

Non-genetic health care professionals lack of awareness of patient risk factors and potential genetic component.

Failure to obtain an adequate family history
Lack of genetic workforce to have adequate staffing to enable program enhancements and initiatives.
BARRIERS - PARENTS AND FAMILIES

- Cost of genetic testing/counseling
- Lack of awareness of personal risk
- Unclear information regarding family history
- Access to appropriate care (distance/wait time)
- Delayed diagnosis
- Limited or non-existing treatments
JULIE HOWARD

Expecting the Unexpected: Prematurity and Rare Malformations

TRANSFORMING GOOD SCIENCE INTO GREAT MEDICINE FOR RARE GENETIC DISEASES.
Learning how to deal with a rare malformation, and learning how to pronounce it.
Rhombencephalosynapsis (RES)

Rhombencephalosynapsis is a midline brain malformation characterized by missing cerebellar vermis with apparent fusion of the cerebellar hemispheres.
Dr. Google
Don’t let it happen!

Give information when giving a diagnosis.
What it looks like for Spencer:

- *He is behind physically and ahead mentally which is a hard combination. He requested a brain transplant when he was 8.
- *His neurologist jokes that Spencer will be his college lab assistant in 9th grade.
- *Wish I had a crystal ball.
When possible, connect families new to a diagnosis with other families with the same rare condition.
Inequities and Disparities in Treatment
What can Clinicians and Families Do?
Promote education on rare diseases and malformations for the provision of care to infant and family.
Learn where to find both the medical and developmental resources that the infant will need.
 Advocate for comprehensive newborn screening.
Advocate for resources for families.
IN SUMMARY:

• Educate yourself on rare diseases and malformations and how they affect infants and their families.
• Become competent in identifying, advocating for, and obtaining the services and resources that infants and their families need.
• Build an educated and experienced team of providers and peers you can turn to.
• Stay informed of the emerging pharmacologic treatments for some rare diseases.