

# Rare Disease Fair & Summit

Fair: Friday, May 5, 2023  
8:30 a.m. to 5 p.m.

## Join us for the Rare Disease Fair at:

Seattle Children's Research Institute  
Building Cure 1920 Terry Ave.  
Seattle, WA 98101

To register visit our [website](#)

Virtual Option

[Join link](#)

Webinar code: 2452 616 5994

Password: rarefair (72733247 from phones or video)

**All times are listed in Pacific time**

8:30 a.m.	<b>Introduction</b> Brittany Richey, <i>Manager, Administration, Seattle Children's Research Institute</i>
8:35 a.m.	<b>Welcome</b> Carolina Sommer, <i>Founder and CEO, Born a Hero Research Foundation, Co-Founder of NW Rare Disease Coalition</i>
8:45 a.m.	<b>Introduction</b> Malachi Castillote, <i>Patient Advocate and Rare Artist</i>
<b>Neurological Rare Diseases</b>	
	<b>Moderator</b> Ellen Morgan, <i>Founder and President of Pros Foundations</i>
8:50 a.m.	<b>Brain Organoids in the Service of CTNNB1 Syndrome</b> Dr. Rami Aqeilan, <i>Chair, Institute for Medical Research Israel-Canada (IMRIC) Lautenberg Center for Immunology &amp; Cancer Research Faculty of Medicine, Hebrew University of Jerusalem</i>
9:10 a.m.	<b>CLN2 Disease: An Inborn Error of Metabolism resulting in Neurodegeneration</b> Guillermo Seratti, <i>BioMarin Pharmaceutical</i>
9:30 a.m.	<b>Financial Crisis Rare Diseases Can Put a Family Into</b> Sabrina Castillote, <i>Mom of two with rare Batten's Disease: CLN2 patients, advocate, and caregiver</i>

9:40 a.m.	<p><b>Neurological Overgrowth Rare Disease Panel</b></p> <p><u>Moderator:</u> Ellen Morgan</p> <p><u>Panelists:</u></p> <ul style="list-style-type: none"> <li>- Shawna Feely, <i>Senior Genetic Counselor, Department of Neurology, Seattle Children's Hospital</i></li> </ul> <p>Overgrowth Syndrome Patient Organizations: The power of collaboration and the patient voice in rare disease communities</p> <ul style="list-style-type: none"> <li>- Kim Venarola, <i>Co-Founder &amp; President, Malan Syndrome Foundation</i></li> <li>- Jill Kiernan, <i>Executive Director, Tatton Brown Rahman Syndrome</i></li> </ul>
10:25 a.m.	<p><b>Neurological Rare Diseases Q&amp;A</b></p> <p><u>Moderator:</u> Ellen Morgan</p> <p><u>Panelists:</u> Dr. Rami Aqeilan, Guillermo Serrati, Sabrina Castillote, Kim Venarola, Jill Kiernan, <i>Shawna Feely</i></p>
10:35 a.m.	<b>Break</b> (15 minutes)
10:50 a.m.	<p><b>In His Owen Time</b></p> <p>Lindsey Topping-Schuetz, <i>Mom, Advocate and NW Rare Disease Coalition Board Member</i></p>
11:00 a.m.	<p><b>How My Mom's Balance Disorder Affects Me</b></p> <p>Boston Newell, <i>Young Advocate</i></p>
<b>Investing in Industry and Biotech</b>	
	<p><b>Moderator</b></p> <p>Joshua Henderson, <i>Co-Founder, NW Rare Disease Coalition and Head of Rare Diseases, Pulse Inframe</i></p>
11:05 a.m.	<p><b>The Power of the Patient Voice in Drug Development</b></p> <p><u>Moderator:</u> Joshua Henderson</p> <p><u>Panelists:</u></p> <ul style="list-style-type: none"> <li>- Wendy Erler, VP Patient Experience: Patient Advocacy and Patient Insights at Alexion <i>Pharmaceuticals</i></li> <li>- Holly Snyder, <i>Senior Manager, Medical Affairs at Illumina</i></li> <li>- Emily Radomile, <i>Vice President, Program Management and Clinical Operations at Mahzi Therapeutics</i></li> </ul>
11:45 a.m.	<p><b>The Policy Levers to Accelerate Access to Care and Research for the Rare Community</b></p> <p><u>Moderator:</u> Joshua Henderson</p> <p><u>Panelists:</u></p> <ul style="list-style-type: none"> <li>- Dr. Marianne Hamilton-Lopez, <i>Senior Research Director, Biomedical Innovation at Duke-Margolis Center for Health Policy</i></li> <li>- Julie Boyd, <i>Director State Government Affairs at Ultragenyx Pharmaceuticals, Inc</i></li> </ul>

12:15 p.m.	<p><b>The Challenges Bringing Cell and Gene Therapies from Lab to Bedside</b></p> <p><u>Moderator:</u> Joshua Henderson</p> <p><u>Panelists:</u></p> <ul style="list-style-type: none"> <li>- Dr. Jeff Chamberlain, <i>Professor in Departments of Neurology, Medicine, and Biochemistry, University of Washington School of Medicine, McCaw Endowed Chair in Muscular Dystrophy</i></li> <li>- Shelly Eagen, <i>US Medical Affairs, Rare Disease Neurology at Pfizer</i></li> </ul>
12:45 p.m.	<p><b>Policy Discussion</b></p> <p><u>Moderator:</u> Max Brown</p> <p><u>Panelists:</u></p> <ul style="list-style-type: none"> <li>- Marc Cummings, <i>President &amp; CEO of Life Science Washington</i></li> <li>- Katie Stoll, <i>Executive Director at Genetic Support Foundation</i></li> </ul>
1:05 p.m.	<b>Lunch</b> (45 minutes)
1:50 p.m.	<p><b>Our Journey</b></p> <p>Effie Parks, Host of Once Upon A Gene Podcast</p>
2:00 p.m.	<p><b>Singing 'Tomorrow'</b></p> <p>Esme Parks, <i>Sister-Advocate of CTNN1B</i></p>
<b>Policy</b>	
	<p><b>Moderator</b></p> <p>Max Brown, <i>Co-Founder, NW Rare Disease Coalition, Vice President of Public Affairs at Desimone Consulting Group</i></p>
2:05 p.m.	<p><b>Washington Rare Disease Advisory Council Model</b></p> <p><u>Moderator:</u> Max Brown, Lindsey Topping-Schuetz</p> <p>Video introduction – State Representative My-Linh Thai</p> <p><u>Panelist:</u></p> <ul style="list-style-type: none"> <li>- Mary McDirmid, <i>NW Rare Disease Coalition Board Member, Founder of Special Abilities Network</i></li> </ul>
2:30 p.m.	<p><b>Whole Genome Sequencing Bill Panel</b></p> <p><u>Moderator:</u> Max Brown, Sarah Tompkins</p> <p><u>Panelists:</u></p> <ul style="list-style-type: none"> <li>- Jessie Conta, <i>Laboratory Stewardship Consultant, PLUGS® &amp; Pickhandle Consulting</i></li> <li>- Madison Arenchild, <i>Manager, Market Access at Rady Children's Institute for Genomic Medicine</i></li> </ul>
2:55 p.m.	<b>Break</b> (15 minutes)

3:10 p.m.	<b>Introduction</b> Karen Pratt and her two warriors Luke, and Jack Pratt
<b>Genetic Testing</b>	
	<b>Moderator</b> Joshua Henderson, <i>Co-Founder, NW Rare Disease Coalition and Head of Rare Diseases, Pulse Inframe</i>
3:15 p.m.	<b>Diagnosis Now, Treatment Later, and Rare Point of View</b> Janie Reade, <i>Author, Parent Coach</i>
3:25 p.m.	<b>Gaps in Genetic Testing for Rare Diseases</b>  <u>Moderators:</u> Holly Snyder  <ul style="list-style-type: none"> <li>- Jessie Conta, <i>Laboratory Stewardship Consultant, PLUGS® &amp; Pickhandle Consulting</i></li> <li>- Charlotte Skinner, <i>Genetic Counselor and Data Analyst at Concert Genetics</i></li> <li>- Janie Reade, <i>Author, Life Coach, Parent Advocate</i></li> <li>- Katie Golden-Grant, <i>Senior Genetic Counselor at Illumina, Lecturer at University of Washington</i></li> <li>- Claudia Carvalho, <i>Assistant Investigator at PNRI</i></li> </ul>
4:10 p.m.	<b>Time is of the Essence</b> Corrin Jackson, <i>Batten Disease Parent Advocate</i>
4:20 p.m.	<b>Introduction</b> Owen and his cousin
<b>NW Rare Disease Coalition Task Forces</b>	
4:25 p.m.	<b>Mental Health Task Force</b> <ul style="list-style-type: none"> <li>- Carolina Sommer, <i>Co-Founder of NW Rare Disease Coalition, CEO/Founder of Born a Hero Research Foundation</i></li> <li>- Kari Cunningham-Rosvik, <i>Co-Chair, Mental Health Task Force, Parent Advocate</i></li> <li>- Christina Newell, <i>Licensed Mental Health Counselor, Former Art Teacher, and Rare Disease Patient</i></li> </ul>
4:33 p.m.	<b>Genetic Testing Task Force</b> <ul style="list-style-type: none"> <li>- Carolina Sommer, <i>Co-Founder of NW Rare Disease Coalition, CEO/Founder of Born a Hero Research Foundation</i></li> <li>- Jessie Conta, <i>Laboratory Stewardship Consultant, PLUGS® &amp; Pickhandle Consulting</i></li> </ul>
4:41 p.m.	<b>Incidence and Prevalence Task Force</b> <ul style="list-style-type: none"> <li>- Carolina Sommer, <i>Co-Founder of NW Rare Disease Coalition, CEO/Founder of Born a Hero Research Foundation</i></li> <li>- Mary McDirmid, <i>Chair, Incidence and Prevalence Task Force, Founder, Special Abilities Network, and Parent Advocate</i></li> </ul>
4:46 p.m.	<b>Rural Rare Disease Community Project Task Force</b> <ul style="list-style-type: none"> <li>- Carolina Sommer, <i>Co-Founder of NW Rare Disease Coalition, CEO/Founder of Born a Hero Research Foundation</i></li> <li>- Marissa Dunn, <i>Regional Director Advocacy, Vertex Pharmaceuticals</i></li> </ul>
4:55 p.m.	<b>Closing Remarks</b> Joshua Henderson, <i>Co-Founder, NW Rare Disease Coalition, Head of Rare Diseases, Pulse Inframe</i>
5:00 p.m.	<i>Adjourn</i>

# Rare Disease Fair & Summit

Summit: Saturday, May 6, 2023  
8:30 a.m. to 1:40 p.m.

Click [HERE](#) to Join the Rare Disease Summit:

**Meeting Code:** 2466 687 3434

**Password:** raresummit

**All times are listed in Pacific time**

8:30 a.m.	<b>Introduction</b> Brittany Richey, <i>Manager, Administration, Seattle Children's Research Institute</i>
8:35 a.m.	<b>Welcome</b> Joshua Henderson, <i>Co-Founder, NW Rare Disease Coalition and Head of Rare Diseases, Pulse Inframe</i>

## Challenges Faced by Patients: Real Stories and Advice

	<b>Moderator</b> Carolina Sommer, <i>Founder and CEO, Born a Hero Research Foundation, Co-Founder of NW Rare Disease Coalition</i>
8:45 a.m.	<b>Patient Perspective: Anxiety, Depression, Trauma...and Tools</b> Kara Jackman, <i>CCA Adult Programs Coordinator</i>
9:10 a.m.	<b>Mental Health Impact on Rare Disease Loved Ones</b> Allison Fine, <i>Executive Director, The Center for Chronic Illness</i>

## Policy 101

	<b>Moderator</b> Sarah Tompkins, <i>Classical Ehlers-Danlos Syndrome &amp; Rare Diseases Patient Advocate, NW Rare Disease Board Member, Eastside EDS Board Member, Ms. Wheelchair USA Ambassador</i>
9:35 a.m.	<b>Rare Disease Stories for Understanding</b> Kathy Devanny, <i>Director of Advocacy and HCP Relations, Patient Worthy</i>
9:55 a.m.	<b>Policy &amp; Advocacy 101</b> Max Brown, <i>Co-Founder NW Rare Disease Coalition, Vice President of Public Affairs at Desimone Consulting Group</i> Emma Scalzo, <i>Alliance Development Manager, Desimone Consulting Group</i>
10:20 a.m.	<b>Break</b> (15 minutes)

## Care Management

	<b>Moderator</b> Lindsey Topping Schuetz, <i>NW Rare Disease Coalition Board Member</i>
10:35 a.m.	<b>Putting Me Back in Medicine</b> Ryan Sheedy, <i>Founder of Mejo</i>
11:00 a.m.	<b>Self-Care for Your Healthcare</b> Sarah Tompkins, <i>NW Rare Disease Coalition, Eastside EDS Board Member, EDS, and Rare Diseases Patient Advocate</i>
11:20 a.m.	<b>How I became a better parent to my child with extreme special needs by lifting my emotional burdens</b> Janie Reade, <i>Author, Life Coach, Rare Disease Parent</i>
11:40 a.m.	<b>Making Art: Self-Compassion, Mindfulness, and Healing</b> Christina Newell, <i>Licensed Mental Health Counselor, Former Art Teacher, and Rare Disease Patient</i>
12:00 p.m.	<b>Break</b> (15 minutes)
12:15 p.m.	<b>Tiny Habits - How to Get Done What You've Always Wanted to Do</b> David Sommer, <i>Vice President, Born a Hero. Associate Principal, Degenkolb Engineers</i>
12:45 p.m.	<b>An Introduction to Informing Families and Life Course Tools Which Can Help You and Your Family Explore and Design Your Vision of a Good Life</b> Krista Perleberg, <i>Family Coordinator for Informingfamilies.org</i>
1:10 p.m.	<b>Extra Layers of Support: Complex Care Coordination and Palliative Care</b> Michele Hagerstrom, <i>Complex Primary Care, Palliative Care, and Physical Medicine &amp; Rehab clinics at Mary Bridge Children's</i>
1:30 p.m.	<b>Closing Remarks</b> Carolina Sommer, <i>Founder and CEO, Born a Hero Research Foundation, Co-Founder of NW Rare Disease Coalition</i>
1:40 p.m.	<b>END</b>

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