

<b>Agenda</b>	
<b>Solve-RD Annual Meeting 2020</b>	
Date	5-6 March 2020
Venue	Parc Científic de Barcelona (PCB) Calle Baldori i Reixac, nº4 Tower I and D 08028 Barcelona, Spain

### Wednesday, 4 March – satellite meeting (invitation only) –

<b>15-18</b>	<b>CETF Meeting</b> <i>Chair: Gulcin Gumus and Virginie Bros-Facer, EURORDIS</i>
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### Thursday, 5 March

<b>8:30</b>	<b>Registration</b> ( <i>foyer, Tower D, ground floor</i> )			
<b>9:00</b>	<b>DITF meeting: joint session</b> ( <i>Chair: Alex Hoischen and Sergi Beltran; Room: auditorium, Tower D, ground floor</i> ) <ul style="list-style-type: none"> <li>Updates and plans to enable discussion in the DITF sessions</li> </ul>			
<b>9:45</b>	<b>DITF meeting: break-out sessions</b> ( <i>Chair: DITF leads</i> )			
	<b>ITHACA</b> ( <i>Chair: Lisenka Vissers; Room: auditorium, Tower D</i> )	<b>RND</b> ( <i>Chair: Rebeca Schüle and Matthias Synofzik; Room: sala 1, Tower I</i> )	<b>EURO-NMD</b> ( <i>Chair: Ana Topf; Room: sala 3, Tower I</i> )	<b>GENTURIS</b> ( <i>Chair: Richarda de Voer; Room: sala 2, Tower I</i> )
<b>11:15</b>	<b>Coffee break</b> ( <i>foyer, Tower D, ground floor</i> )			
<b>11:45</b>	<b>DITF meeting: break-out sessions – continued</b> ( <i>Chair: DITF leads</i> )			
	<b>ITHACA</b> ( <i>Chair: Lisenka Vissers; Room: auditorium, Tower D</i> )	<b>RND</b> ( <i>Chair: Rebeca Schüle and Matthias Synofzik; Room: sala 1, Tower I</i> )	<b>EURO-NMD</b> ( <i>Chair: Ana Topf; Room: sala 3, Tower I</i> )	<b>GENTURIS</b> ( <i>Chair: Richarda de Voer; Room: sala 2, Tower I</i> )
<b>13:15</b>	<b>Lunch</b> ( <i>foyer, Tower D, ground floor</i> ) <b>Registration</b> ( <i>foyer, Tower D, ground floor</i> )			
<b>14:30</b>	<b>Opening session</b> ( <i>Chair: Olaf Riess; Room: auditorium, Tower D, ground floor</i> ) <ul style="list-style-type: none"> <li>Welcome (<i>Olaf Riess, EKUT and Sergi Beltran, CNAG-CRG – 10 min</i>)</li> </ul>			
<b>14:40</b>	<b>Policies &amp; project logistics</b> ( <i>Chair: Holm Graessner, Room: auditorium, Tower D, ground floor</i> )			
<b>15:00</b>	<b>WP1 – unique RD patients and phenotypes</b> ( <i>Chair: Ana Rath; Room: auditorium, Tower D, ground floor</i> )			

	<ul style="list-style-type: none"> <li>Data collection (<i>Sergi Beltran, CNAG-CRG – 10+5 min</i>)</li> <li>PhenoPackets (<i>Peter Robinson, JAX – 10+5 min</i>)</li> <li>RDCO ontology (<i>Ana Rath &amp; David Lagorce, INSERM-Orphanet – 10+5 min</i>)</li> </ul>		
<b>15:45</b>	<b>Coffee break &amp; Poster session</b> ( <i>foyer, Tower D, ground floor</i> ) All poster will be presented		
<b>16:45</b>	<table border="1"> <tr> <td> <p><b>WP3 – changing patients’ lives</b> (<i>Chair: Gisèle Bonne; Room: auditorium, Tower D, ground floor</i>)</p> <ul style="list-style-type: none"> <li>Improving the communication of genomics results using Experience-Based Co-Design – Process and outcomes (<i>Alessia Costa, KCL and Vera Frankova, CUP – 15+5 min</i>)</li> <li>Assessing Cost-Effectiveness and Clinical Utility of WES/WGS - Progress report (<i>Camille Level, University of Burgundy – 10+5 min</i>)</li> <li>Treatabome (<i>Gisèle Bonne, INSERM-CRM – 10+5 min</i>)</li> <li>Winter School: capacity building programme for patients on scientific innovation and translational research (<i>Virginie Bros-Facer, EURORDIS – 8+2 min</i>)</li> </ul> </td> <td> <p><b>DATF meeting</b> (<i>Chair: Alex Hoischen and Sergi Beltran; Room: sala 3, Tower I</i>)</p> </td> </tr> </table>	<p><b>WP3 – changing patients’ lives</b> (<i>Chair: Gisèle Bonne; Room: auditorium, Tower D, ground floor</i>)</p> <ul style="list-style-type: none"> <li>Improving the communication of genomics results using Experience-Based Co-Design – Process and outcomes (<i>Alessia Costa, KCL and Vera Frankova, CUP – 15+5 min</i>)</li> <li>Assessing Cost-Effectiveness and Clinical Utility of WES/WGS - Progress report (<i>Camille Level, University of Burgundy – 10+5 min</i>)</li> <li>Treatabome (<i>Gisèle Bonne, INSERM-CRM – 10+5 min</i>)</li> <li>Winter School: capacity building programme for patients on scientific innovation and translational research (<i>Virginie Bros-Facer, EURORDIS – 8+2 min</i>)</li> </ul>	<p><b>DATF meeting</b> (<i>Chair: Alex Hoischen and Sergi Beltran; Room: sala 3, Tower I</i>)</p>
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<b>17:45</b>	<p><b>WP5 – impact and engagement</b> (<i>Chair: Gulcin Gumus</i>)</p> <ul style="list-style-type: none"> <li>Update on the development of WGS diagnostic guidelines (<i>Holm Graessner, EKUT –5 min</i>)</li> <li>Community Engagement Task Force (<i>Gulcin Gumus, EURORDIS – 10+5 min</i>)</li> </ul>		
<b>18:05</b>	<b>On own</b>		
<b>20:00</b>	<b>Joint dinner at restaurant Alba Granados</b> ( <i>C/ Enric Granados 34, 08036 Barcelona; <a href="http://www.albagranados.cat">www.albagranados.cat</a></i> )		

### Friday, 6 March (*auditorium, Tower D, ground floor*)

<b>8:30</b>	<p><b>WP2 – novel molecular causes</b> (<i>Chairs: Alex Hoischen and Sergi Beltran</i>)</p> <ul style="list-style-type: none"> <li>Results re-analysis / cohort 1 (<i>Sergi Beltran, CNAG-CRG – 10+5 min</i>)</li> <li>CNV analysis: results &amp; data interpretation (<i>Steve Laurie, CNAG-CRG – 15+5 min</i>)</li> <li>SNV/indel analysis: results &amp; data interpretation (<i>Leslie Matalonga, CNAG-CRG – 15+5 min</i>)</li> <li>Runs of homozygosity and relatedness analysis: results &amp; data interpretation (<i>Julia Schulze-Hentrich, EKUT – 15+5 min</i>)</li> <li><i>De novo</i> trio analysis   Meta-analysis novel genes/gene burden analysis: results &amp; data interpretation (<i>Christian Gilissen, RUMC – 15+5 min</i>)</li> <li>Discussion (20 min)</li> </ul>
<b>10:30</b>	<b>Coffee break</b>
<b>11:00</b>	<p><b>WP2 – novel molecular causes – continued</b> (<i>Chairs: Alex Hoischen and Sergi Beltran</i>)</p> <ul style="list-style-type: none"> <li>Novel omics: plans, timelines, results (<i>Alex Hoischen, RUMC – 5 min</i>)</li> </ul>

	<ul style="list-style-type: none"> <li>▪ Novel omics: cohorts, use cases, results ITHACA (<i>Lisenka Vissers, RUMC – 10 min</i>)</li> <li>▪ Novel omics: cohorts, use cases, results RND (<i>Rebecca Schüle and Matthis Synofzik, EKUT – 10 min</i>)</li> <li>▪ Novel omics: cohorts, use cases, results EURO-NMD (<i>Ana Topf, UNEW – 10 min</i>)</li> <li>▪ Novel omics: cohorts, use cases, results GENTURIS (<i>Richarda de Voer, RUMC – 10 min</i>)</li> <li>▪ Discussion (<i>15 min</i>)</li> </ul>
<b>12:00</b>	Keynote lecture ( <i>Alexandre Reymond, Center for Integrative Genomics, University of Lausanne – 30 min</i> )
<b>12:30</b>	<b>Group photo</b> Lunch ( <i>foyer, Tower D, ground floor</i> )
<b>14:00</b>	<b>WP4 – bioinformatics and knowledge management</b> ( <i>Chair: Anthony Brookes</i> ) <ul style="list-style-type: none"> <li>▪ Data standards (<i>Dylan Spalding, EMBL-EBI – 5+5 min</i>)</li> <li>▪ Discovery: Patient Matchmaking, Datasets, Researchers and Samples (<i>Anthony Brookes, ULEIC – 5+5 min</i>)</li> <li>▪ GPAP services and data sharing (<i>Sergi Beltran, CNAG-CRG – 10+5 min</i>)</li> <li>▪ Sandbox &amp; RD3 database (<i>Morris Swertz, UMCG – 15+10 min</i>)</li> </ul>
<b>15:00</b>	<b>End of meeting</b>
<b>15:00</b>	<b>Sandbox discussion</b> ( <i>Chair: Holm Graessner, room: sala 3, Tower I</i> )
<b>17:00</b>	<b>Solve-RD Steering Committee wrap up</b> ( <i>room: sala 3, Tower I</i> ) Invited: Steering Committee (SC), Scientific Advisory Board (SAB), Independent Ethics Advisory Board (IEAB)
<b>17:30</b>	<b>On own</b>
<b>20:00</b>	<b>Solve-RD SC dinner (optional)</b> (tbd)

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