

Agenda

Solve-RD Annual Meeting 2020

Date	5-6 March 2020
Venue	Parc Científic de Barcelona (PCB) Calle Baldori i Reixac, nº4 Tower I and D 08028 Barcelona, Spain

Thursday, 5 March

8:30	Registration (<i>foyer, Tower D, ground floor</i>)			
9:00	DITF meeting: joint session (<i>Chair: Alex Hoischen; Room: auditorium, Tower D, ground floor</i>)			
	▪			
9:45	DITF meeting: break-out sessions (<i>Chair: DITF leads</i>)			
	ITHACA (<i>Chair: Lisenka Vissers; Room: auditorium, Tower D</i>)	RND (<i>Chair: Rebeca Schüle and Matthias Synofzik; Room: sala 1, Tower I</i>)	EURO-NMD (<i>Chair: Ana Topf; Room: sala 3, Tower I</i>)	GENTURIS (<i>Chair: Richarda de Voer; Room: sala 2, Tower I</i>)
11:15	Coffee break (<i>foyer, Tower D, ground floor</i>)			
11:45	DITF meeting: break-out sessions – continued (<i>Chair: DITF leads</i>)			
	ITHACA (<i>Chair: Lisenka Vissers; Room: auditorium, Tower D</i>)	RND (<i>Chair: Rebeca Schüle and Matthias Synofzik; Room: sala 1, Tower I</i>)	EURO-NMD (<i>Chair: Ana Topf; Room: sala 3, Tower I</i>)	GENTURIS (<i>Chair: Richarda de Voer; Room: sala 2, Tower I</i>)
13:15	Lunch (<i>foyer, Tower D, ground floor</i>)			
	Registration (<i>foyer, Tower D, ground floor</i>)			
14:15	Opening session (<i>Chair: Holm Graessner; Room: auditorium, Tower D, ground floor</i>)			
	<ul style="list-style-type: none"> ▪ Welcome (<i>Olaf Riess, EKUT and Sergi Beltran, CNAG-CRG – 10 min</i>) ▪ Keynote lecture (<i>td, tbd – 35 min</i>) 			
15:00	Policies & project logistics (<i>Chair: Holm Graessner; Room: auditorium, Tower D, ground floor</i>)			
15:20	WP1 – unique RD patients and phenotypes (<i>Chair: Ana Rath; Room: auditorium, Tower D, ground floor</i>)			
	<ul style="list-style-type: none"> ▪ Data collection (<i>tbd, CNAG-CRG – 10+5 min</i>) ▪ RDCO ontology (<i>tbd, INSERM-Orphanet – 10+5 min</i>) ▪ PhenoPackets (<i>tbd, JAX – 10+5 min</i>) 			
16:05	Coffee break & Poster session (<i>foyer, Tower D, ground floor</i>)			
	Poster with even number s will be presented			
17:00	WP3 – changing patients' lives (<i>Chair: Gisèle Bonne; Room: auditorium, Tower D, ground floor</i>)			

	<ul style="list-style-type: none"> EBCD study (<i>Alessia Costa, KCL and Alison Metcalfe, SHU – 15+5 min</i>) Systematic review (<i>Aurore Pélissier, University of Burgundy – 10+5 min</i>) Treataboloome (<i>Gisèle Bonne, INSERM-CRM – 10+5 min</i>) Winter School (<i>Virginie Bros-Facer, EURORDIS – 8+2 min</i>) 	
18:00	Ethics session (<i>Chair: Solve-RD IEAB; Room: tbd</i>)	DATF Meeting (<i>Chair: Sergi Beltran; Room: tbd</i>)
19:00	On own	
20:00	Joint dinner at restaurant Citrus (Passeig de Gràcia, 44, 08007 Barcelona; www.citrusrestaurant.es)	

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

9:00	WP2 – novel molecular causes (<i>Chairs: Alex Hoischen and Sergi Beltran</i>) <ul style="list-style-type: none"> Results re-analysis / cohort 1 (<i>Sergi Beltran, CNAG-CRG – 10+5 min</i>) CNV analysis: results & data interpretation (<i>Steve Laurie, CNAG-CRG – 15+5 min</i>) SNV/indel analysis: results & data interpretation (<i>Leslie Matalonga, CNAG-CRG – 15+5 min</i>) Runs of homozygosity and relatedness analysis: results & data interpretation (<i>Stephan Ossowski, EKUT – 15+5 min</i>) De novo trio analysis: results & data interpretation (<i>Christian Gilissen, RUMC – 15+5 min</i>) Meta-analysis novel genes/gene burden analysis: results & data interpretation (<i>Christian Gilissen, RUMC – 15+5 min</i>) 	
11:00	Coffee break	
11:30	WP2 – novel molecular causes – continued (<i>Chairs: Alex Hoischen and Sergi Beltran</i>) <ul style="list-style-type: none"> Novel omics: plans, timelines, results (<i>Alex Hoischen, RUMC – 5 min</i>) Novel omics: cohorts, use cases, results ITHACA (<i>Lisenka Vissers, RUMC – 10 min</i>) Novel omics: cohorts, use cases, results RND (<i>Rebecca Schüle and Matthis Synofzik, EKUT – 10 min</i>) Novel omics: cohorts, use cases, results EURO-NMD (<i>Ana Topf, UNEW – 10 min</i>) Novel omics: cohorts, use cases, results GENTURIS (<i>Richarda de Voer, RUMC – 10 min</i>) Discussion (<i>15 min</i>) 	
12:30	Group photo Lunch (<i>foyer, Tower D, ground floor</i>)	
13:30	Poster session (<i>foyer, Tower D, ground floor</i>) Poster with odd numbers will be presented	
14:30	WP4 – bioinformatics and knowledge management (<i>Chair: Anthony Brookes</i>) <ul style="list-style-type: none"> Sandbox & RD3 database (<i>Morris Swertz, UMCG – 10+5 min</i>) GPAP services and data sharing (<i>Sergi Beltran, CNAG-CRG – 10+5 min</i>) Discovery: Patient Matchmaking, Datasets, Researchers and Samples (<i>Anthony Brookes, ULEIC – 10+5 min</i>) Data standards (<i>Dylan Spalding, EMBL-EBI – 10+5 min</i>) 	

15:30	WP5 – impact and engagement (<i>Chair: Gulcin Gumus</i>) <ul style="list-style-type: none"> ▪ Update on the development of WGS diagnostic guidelines (<i>Hans Scheffer, RUMC – 10+5 min</i>) ▪ Community Engagement Task Force (<i>Gulcin Gumus, EURORDIS – 10+5 min</i>)
16:00	End of meeting
17:00	Solve-RD Steering Committee meeting (<i>sala 1, Tower I</i>) Invited: Steering Committee (SC), Scientific Advisory Board (SAB), Independent Ethics Advisory Board (IEAB)
19:00	On own
20:00	Solve-RD SC dinner (optional) (tbd)

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