5<sup>th</sup> Training course on "Quality assurance, variant interpretation and data management in the NGS diagnostics era"

18-20 Oct 2023, Warsaw, Poland

PROGRAM



# **European Joint Programme on Rare Diseases**

H2020-SC1-2018-Single-Stage-RTD

SC1-BHC-04-2018

Rare Disease European Joint Programme Cofund



Grant agreement number 825575

5<sup>th</sup> Training Course on

"Quality assurance, variant interpretation and data management

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## Day 1 – 18 October 2023

		Speaker	Affiliation
	Arrivals		
11:00-	Registration Desk		
13:00			
12:00	Lunch		
12.00			
13:00	Opening & Welcome	Krystyna Chrzanowska	The Children's Memorial Health Institute, Warsaw, Poland
Session 2	1 - NGS diagnostics		
13:15	The importance of NGS in daily clinical practice: the impact of a	Saskia	Paracelsus Medical University,
	genetic diagnosis for treatment and beyond	Wortmann	Salzburg, Austria
14:15	Efficient analysis of whole genome sequencing data	Tomasz	University of Bergen, Norway
		Stokowy	
15:15	Dataspace and databases. Legal aspects, existing and emerging	Mahsa	University of Ghent, Belgium
	regulatory frameworks (GDPR, European Health Data Space)	Shabani	
	and use of federated networks in pan-European genomic		
	projects.		
16:00	Coffee Break		
Socion	2 - Hands-on: Variant filtering and prioritisation	Leslie	CNAG-CRG, Barcelona, Spain
363510117	2 - Hanus-on. Variant Intering and promisation	Matalongo,	CNAG-CKG, Barcelona, Span
		Steven Laurie	
16:30	Short introduction on data filtering, prioritisation and		
	classification + GPAP		
	Case demo introducing key concepts		
	Explanation and use of variant classification following		
10.00	ACMG guidelines		
18:00	End Day 1		

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		Speaker	Affiliation
Session	3 - Quality in NGS diagnostics		
9:00	Diagnostic Laboratory quality assurance and management	Gert Matthijs	KU Leuven, Belgium
9:30	The Polish experience on the external quality assurance of	Mateusz	Institute of Mother and
	genetic testing.	Dawidziuk	Child, Warsaw, Poland
10:00	Recommendations for whole genome sequencing in diagnostics for rare diseases	Erika Souche	UZ/KU Leuven, Belgium
10:30	Coffee Break		
Session	4 – Clinical and functional validation		
11:00	The importance of (reverse) phenotyping, communication and functional validation for variant interpretation	Saskia Wortmann	Paracelsus Medical University, Salzburg, Austria
11:45	Multi OMICS data in Mendelian disease diagnostics	Holger Prokisch	Technical University of Munich, Germany
12:30	Lunch		
Session	5 – Innovation and research		
13:30	Universal PGT: achievements and challenges	Eftychia Dimitriadou	UZ/KU Leuven, Belgium
14:15	How exome sequencing changes the prenatal diagnostics	Beata Nowakowska	Institute of Mother and Child, Warsaw, Poland
15:00	Follow-up of rare autosomal trisomies revealed by NIPT	Eftychia Dimitriadou	UZ/KU Leuven, Belgium
15:45	Coffee Break		
Session 6 - Hands-on (part 2) - Variant filtering and prioritisation		Leslie Matalongo, Steven Laurie	CNAG-CRG, Barcelona, Spain
16:15	Background on ACMG classification and tools for correct interpretation		
	Hands-on solving cases		
	Q&A		
17:30	End Day 2		

## Day 2 - 19 October 2023

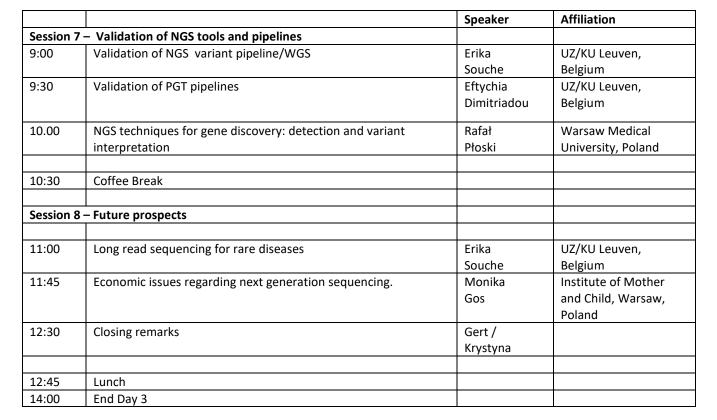




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## Day 3 - 20 October 2023



EUROPEAN SOCIETY OF HUMANA GENETICS