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5th Training Course on

"Quality assurance, variant interpretation and data management

in the NGS diagnostics era"

[18-20 Oct 2023]

Warsaw, Poland

Pillar 3 - WP14.2 - Standards and quality
of genetics/genomics data in laboratory and clinical practice

5th Training course on
"Quality assurance, variant interpretation and data management
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18-20 Oct 2023, Warsaw, Poland

PROGRAM

Day 1 – 18 October 2023

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

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Day 2 – 19 October 2023

		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 genetic testing.	Mateusz Dawidziuk	Institute of Mother and 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		

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Day 3 – 20 October 2023

		Speaker	Affiliation
Session 7 – Validation of NGS tools and pipelines			
9:00	Validation of NGS variant pipeline/WGS	Erika Souche	UZ/KU Leuven, Belgium
9:30	Validation of PGT pipelines	Eftychia Dimitriadou	UZ/KU Leuven, Belgium
10.00	NGS techniques for gene discovery: detection and variant interpretation	Rafał Płoski	Warsaw Medical University, Poland
10:30	Coffee Break		
Session 8 – Future prospects			
11:00	Long read sequencing for rare diseases	Erika Souche	UZ/KU Leuven, Belgium
11:45	Economic issues regarding next generation sequencing.	Monika Gos	Institute of Mother and Child, Warsaw, Poland
12:30	Closing remarks	Gert / Krystyna	
12:45	Lunch		
14:00	End Day 3		