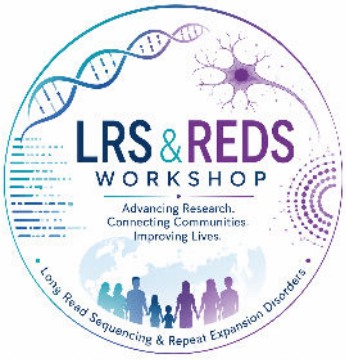

Long-Read Sequencing of Expanded Tandem repeats
Workshop 3

Wednesday 23th September to Thursday 24th September
Paris, FRANCE

OPENMIND (Close to OPERA GARNIER)
3 Boulevard des Capucines, 75002 Paris
(Subway lines 3, 7, 8, 9 and RER A and E)

Wednesday 23th	
8:30-9:00	Arrival and Welcome
Session I: Repeat expansion analyses- New Advances	
9:00-9:30 (30min)	***Adam Ameur (Uppsala, Sweden)*** Keynote
9:30-11:00 (20min)	***Dusaka Savic-Pavcevic (University of Belgrade)*** Longitudinal Dynamics of Somatic Expansion in Blood Reflects Muscle Progression in Myotonic Dystrophy Type (SP-PCR) ***Nehaa Kandasamy Packil Ponraj (University of Glasgow)*** Small pool PCR pooled ONT sequencing of DM1 patient fibroblast cells over time ***Eva Coll (IGTP, Barcelona)*** Comparison between long-read sequencing technologies and the gold-standard Small Pool PCR in the study of Myotonic Dystrophy Type 1 ***Deborah Moine and Tom Mokveld (PacBio)*** PureTarget for Challenging Genomic Regions & Repeat Expansions: Workflow & Analysis
11:00-11:30	Discussion and Coffee break
Session II: No-amplification targeted approaches	
11:30-13:00 (20min)	***Vitus Prokosh (MGZ)*** Applications and Performance of Cas9-Enriched Long-Read Sequencing with ONT and PacBio for Short Tandem Repeat Analysis ***Corentin Schwarz, Natalia Pinzón and Stéphanie Tomé (Strasbourg Hospital, Myology institute and Center of Myology, INSERM)*** Long-read sequencing of complex myotonic dystrophy type 1 cases reveals CCG-enriched expanded alleles (ONT and PacBio)

	Djihah Hadjadj (Cochin Institute, Paric Cité université) PacBio CiFi long-read multiomics to profile DNA methylation, 3D genome organization and genetic variation ***Thomas Hoekman (Radboudumc)*** Single molecule analysis reveals asymmetric CpG methylation at the DM1 locus
13:00-14:30	Lunch
Session IIIA: New discoveries through amplification-free targeted sequencing	
14:30-16:00 (20min)	***Birgitt Schuele (Standford University)*** Beyond Repeat Length: Defining How Alternate Motifs Drive SCA10 Pathogenesis ***Jovan Pesovic (University of Belgrade)*** Precise characterization of FGF14 repeat expansions for affordable SCA27B diagnostics using amplicon-based nanopore sequencing **Alan Mejía maza, Massachusetts General Hospital** Deciphering the epigenetic mechanisms in carriers of X-linked dystonia parkinsonism (Zoom) ***Indhu Shree Rajan Babu (University of British Columbia)*** Long-Read Sequencing and Tandem Repeat Variation in Neurodevelopmental and Psychiatric Disorders (Zoom)
16:00-17:30	Discussion and Coffee
20:30	**Dinner Cruise** Pont Alexandre III behind the Bistrot Alexandre Please arrive on time

Thursday 24th	
9:00	Arrival
Session IIIB: New discoveries through amplification-free targeted sequencing	
9:30-11:00 (20min)	***Marzia Rossato (University of Verona)*** Comparative long-read sequencing of CNBP microsatellites expansions validates novel repeat features in Myotonic Dystrophy Type 2 ***Christina Zarouchlioti (*UCL Institute of Ophthalmology)** Exploring unstable short tandem repeats in the cornea using PureTarget HiFi sequencing ***Jian Chen (CHDI)*** Application of Long Read Sequencing in Huntington's Disease ***Chen Zhongbo (UCL)*** PureTarget on repeat expansion in brain tissue
11:00-11:30	Discussion and Coffee break

Session IV: Bioinformatics tools	
11:30-13:00 (20min)	***Louise Benarroch (Center of Myology, INSERM)*** Comparative Analysis of CRISPR/Cas9-targeted Nanopore Sequencing Approaches in OPDM and DM1 ***Vincent Dion (*UK Dementia research Institute at Cardiff University)** Long-read sequencing of expanded repeats: errors and analysis tools ***Ben Liesfeld (Limbus Medical Technologies GmbH)*** Repeat Expansion Diagnostics Using Long Reads: Assay Design and Analytical Challenges **Lana Radenković *(University of Belgrade)*** Long read sequencing of PCR enriched repeat expansions: tool design, development, and analytical challenges
13:00-14:00	Lunch
Session V: Long read sequencing in REDs diagnostics	
14:00-15:30 (20min)	**Zuzanna Gedziorowska (University of Glasgow)** Comprehensive evaluation of MiSeq, ONT, and PacBio for HTT and TCF4 repeat sequencing: from technical limitations to diagnostic feasibility ***Laura Glaser (MGZ)*** Clinical Implementation of PureTarget HiFi Sequencing for Short Tandem Repeat Expansion Diagnostics ***Brian Sperelakis Beedham (Necker Hospital)*** Long-read sequencing applied to preimplantation genetic diagnosis of rare diseases
Session VI: Round table on challenges to overcome	
15:30-17:30	Discussion/conclusion


PacBio


IDT
INTEGRATED DNA TECHNOLOGIES


NEW ENGLAND
Biolabs


CHDI
FOUNDATION


varvis


LoQus23
THERAPEUTICS


AFMTÉLÉTHON
INNOVER POUR GUÉRIR


3XGENETICS