

Journal Club (30mins+30minsQA) (Thu)1400-1500			Progress report (15mins+5minQA) (Fri)1300-1400		
	Presentor	Topic		Presentor	
2/16	PX	High-coverage whole-genome sequencing of the expanded 1000 Genomes Project cohort including 602 trios	2/17	Calvin	HW Pinfei
2/23	Daphne	Major sex differences in allele frequencies for X chromosomal variants in both the 1000 Genomes Project and gnomAD	2/24	郁書	PX PM
3/2	Pinfei	Myelodysplastic Syndrome associated TET2 mutations affect NK cell function and genome methylation	3/3	庭暄	YT
3/9	YT	Comprehensive SMN1 and SMN2 profiling for spinal muscular atrophy analysis using long-read PacBio HiFi sequencing	3/10	俐伶	俐伶 Calvin

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3/16	PM	Structural variant detection in cancer genomes: computational challenges and perspectives for precision oncology	3/17	HW	Pinfei 郁書
3/23	俐伶	Spinal muscular atrophy diagnosis and carrier screening from genome sequencing data	3/24	PX	PM
3/30	PX	預口試	3/31	YT	庭暄
4/6	HW	Analysis of KIR gene variants in The Cancer Genome Atlas and UK Biobank using KIRCLE - BMC Biology	4/7	Calvin	HW Pinfei
4/13	庭暄		4/14	郁書	PM
4/20		Clara	4/21	庭暄	YT PX
4/27	Pinfei	A cross-disorder dosage sensitivity map of the human genome	4/28	俐伶	俐伶 HW
5/4	YT	Mono- and biallelic variant effects on disease at biobank scale	5/5	Calvin	Pinfei 郁書

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5/11		俐伶	Taiwan Biobank: A rich biomedical research database of the Taiwanese population	5/12		PX	PM 庭暄
5/18		PM	Structural variant detection in cancer genomes: computational challenges and perspectives for precision oncology	5/19		YT	自航 自航
5/25		PX	Structural variation across 138,134 samples in the TOPMed consortium	5/26		Calvin	HW Pinfei
6/1		暫停一次		6/2		郁書	PX PM
6/8				6/9		庭暄	YT
6/15		HW	Analysis of KIR gene variants in The Cancer Genome Atlas and UK Biobank using KIRCLE - BMC Biology	6/16		俐伶	俐伶 Calvin
6/22		端午		6/23		端午	端午 端午
6/29		Pinfei		6/30		HW	Pinfei 郁書

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7/6	YT	Whole genome sequencing identifies structural variants contributing to hematologic traits in the NHLBI TOPMed program - PubMed	7/7	PX	PM 庭暄
7/13	俐伶		7/14	YT	自航 自航
7/20	PM	Structural variant detection in cancer genomes: computational challenges and perspectives for precision oncology	7/21	Calvin	HW Pinfei
7/27	HW	Analysis of HLA Variants and Graves' Disease and Its Comorbidities Using a High Resolution Imputation System to Examine Electronic Medical Health Records	7/28	郁書	PX PM
8/3	庭暄		8/4	庭暄	YT
8/10	PX		8/11	俐伶	俐伶 Calvin
8/17			8/18	HW	Pinfei 郁書
8/24	Pinfei		8/25	PX	PM 庭暄

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8/31	YT		9/1	YT	自航 自航

Updated paper:

- [Truvari: refined structural variant comparison preserves allelic diversity](#)
- [Scaling national and international improvement in virtual gene panel curation via a collaborative approach to discordance resolution](#)
- [Pinfei A cross-disorder dosage sensitivity map of the human genome](#)
- [Profiling variable-number tandem repeat variation across populations using repeat-pangenome graphs](#)
- [Points to consider in the detection of germline structural variants using next-generation sequencing: A statement of the American College of Medical Genetics and Genomics \(ACMG\)](#)
- [A joint NCBI and EMBL-EBI transcript set for clinical genomics and research](#)
- [Genomic analyses of 10,376 individuals in the Westlake BioBank for Chinese \(WBBC\) pilot project](#)
- [Personalized genome assembly for accurate cancer somatic mutation discovery using tumor-normal paired reference samples \(Genome Biology 2022\)](#)
- [Structural variant analysis of a cancer reference cell line sample using multiple sequencing technologies \(Genome Biology 2022\)](#)
- [Variant calling and benchmarking in an era of complete human genome sequences \(Nature Reviews Genetics 2023\)](#)