Poster Presentations

M = Poster Session 1: Monday 5th June | T = Poster Session 2: Tuesday 6th June		Presenting Author
POS001-M	Identifying placental eQTLs and determining biological importance of selected SNPs.	Train Kikas
POS002-T	Multiplex SNP panels based on MALDI-TOF mass-spectrometry genotyping for association analysis of cognitive performance and	Vadim Stepanov
POS003-M	Identification of genes harbouring rare variants of moderate to large effect in schizophrenia: a replication study	Julio Rodriguez
POS004-T	Cito-Nuclear Incompatibility As A Risk Factor For Neurodevelopmental Disorders	Javier Gonzalez-Peñas
POS005-M	Sequence level genotyping at the TCF4 CTG repeat associated with Fuchs endothelial corneal dystrophy	Mariam Alkhateeb
POS006-T	Somatic mutations are abundant in focal cortical dysplasia	Vanessa de Almeida
POS007-M	Association between genetic polymorphisms in RANK/RANKL/OPG signaling pathway with developmental enamel defects and dental caries	Erika Küchler
POS008-T	Association between genetic polymorphisms in DEFB1 and in miRNA202 with caries experience in children	Daniela Oliveira
POS009-M	Modeling atorvastatin plasmatic concentrations in healthy volunteers using integrated pharmacogenetics sequencing	Omar Cruz Correa
POS010-T	Mutational screening of desmosomal and non-desmosomal genes in Russian patients with arrhythmogenic right ventricular cardiomyopathy	Anna Shestak
POS011-M	Association with Idiopathic Pulmonary Fibrosis is attributable to common rather than rare variants in 11p15, 14q21, and 17q21	Jose Miguel Lorenzo Salazar
POS012-T	Identification of a novel CASQ2 deletion causing Catecholaminergic polymorphic ventricular tachycardia	Alejandro Blanco-Verea
POS013-M	Evaluation of genotype-phenotype relationship of short NOP56 expansions causing SCA36	María García-Murias
POS014-T	Triple Negatives in Myeloproliferative Neoplasms: is triple negative positive?	Annabel Kearney
POS015-M	Development of cost/effective strategies for genetic diagnosis of Polycystic Kidney Disease (PKD) based on the population mutagenesis	María Lara Besada Cerecedo
POS016-T	Mapping mitochondrial heteroplasmy in a Leydig tumor by laser capture micro-dissection and cycling temperature capillary electrophoresis	Paulo Refinetti
POS017-M	Identification of disease causing variants of Leukocytoclastic Vasculitis in related individuals	Clara Mulhern
POS018-T	Gene Panel testing Improves Genetic Diagnosis in Hereditary Breast / Ovarian Cancer	Ana Blanco
POS019-M	Germline Promoter Hypermethylation Analysis Of Brca1 And Brca2 Genes In Hereditary Breast And Ovarian Cancer Patients	Marta Rodriguez Balada
POS020-T	The Challenge Of Interpreting Heterozygous Carriers In Niemann-Pick Type C Disease	Cristina Castro Fernández
POS021-M	A Customisable Scripting System for Identification and Filtration of Clinically Relevant Genetic Variants in Whole Exome or Large Gene Panel	Robert Smith
POS022-T	Save variant annotation sharing across laboratories	Beat Wolf
POS023-M	HGVS Variant Descriptions for Short Tandem Repeat Structures	Johan T. den Dunnen for Jonathan Vis
POS024-T	Investigating ACMG rules and quantitative methods for TP53 variant classification	Cristina Fortuno
POS025-M	Variant detection, interpretation and reporting using an end-to-end analysis pipeline	Ruth Burton
POS026-T	Creating new national variation databases and their regulatory environments in Japan	Netsuke Yamamoto
POS027-M	A New National Initiative and Data Sharing Approach for Genomic Medicine in Japan	Jusaku Minari
POS028-T	Interpreting the functional effects of variants with integrated platforms from UniProt	Andrew Nightingale
POS029-M	In silico Prediction of Deleterious SNPs in Endocytosis Genes Implicated in Alzheimer's disease	ChongHan Ng

POS030-T	Application of fibre-FISH in the characterization of complex structural rearrangement and variation: an overview	Fengtang Yang
POS031-M	Overcoming the diagnostic challenges in neurological disorders: the role of next-generation sequencing	Sofia Gouveia
POS032-T	Next-generation sequencing meets splicing: a multiplexed minigene splicing assay for exonic variants of OPN1LW and OPN1MW	Elena Buena Atienza
POS033-M	Utility of trio exome sequencing as a first-line diagnostic test for neurodevelopmental disorders	Ana Fernández-Marmiesse
POS034-T	Massive parallel sequencing for universal indications: diagnostic variant detection in a regional genetic center	Elisabeth Maurer
POS035-M	Targeted Next- Generation Sequencing For Molecular Diagnosis Of Ichthyosis	Uxía-Saraiva Esperón-Moldes
POS036-T	Cycling Temperature Capillary Electrophoresis: A quantitative, fast and inexpensive method to detect mutations in mixed populations of human	Paulo Refinetti
POS037-T	Germline mutations in childhood cancer patients suspected of genetic predisposition to cancer - a retrospective analysis	Dianne Sylvester
POS038-M	Dissecting the molecular basis of epileptic disorders in the Iberian Peninsula.	Sofia Gouveia
POS039-M	PGD counselling for variants of unknown significance	Ekaterina Pomerantseva
POS040-T	Comprehensive variant analyses including whole genome sequencing in hereditary colorectal cancer syndromes	Anna Rohlin
POS041-M	Evaluation Of Basic Massive Parallel Sequencing Parameters In Relation To True/False Positivity's Findings Of Rare Variants From Isolated	Radek Vodicka
POS042-T	Potentially pathogenic germline CHEK2 and NOTCH3 variants among multiple early-onset cancer families	Men Dominguez Valentin
POS043-M	Allelic drop-out is a common phenomenon in the PCR-based NGS and emphasizes the importance of cross-validation.	Anna Bukaeva
POS044-T	Challenges of interpreting sequence variants: experience of a molecular laboratory with a panel of epileptic encephalopathy	Susana Sousa
POS045-M	Copy number variation differences between responders and non-responder to anti-TNF drugs in moderate-to-severe psoriasis	María COvejero-Benito
POS046-T	Identifying limiting factors for single nucleotide and copy number variant calling in a targeted sequencing panel	Pilar Cacheiro
POS047-M	PattRec: An easy-to-use CNV detection tool optimized for targeted NGS panels	Iris Roca
POS048-T	Determining the burden of copy number variation in patients with epilepsy	Tania de Araujo