Genome Structure and Function

374T ‡ High-throughput functional genomic assessment of known and novel *HNF1A* missense variants to support clinical diagnostic interpretation. S. Althari.

377T Primary bilateral macronodular adrenocortical hyperplasia due to mutations in *ARMC5*: New mutations in humans and modeling in zebrafish. A.G. Maria.

380T Exome-wide copy number association study for arsenic-induced skin lesions: A prospective study. M.G. Kibriya.

383T Age-related structural changes of the olfactory receptor subgenome in human blood cells and autologous brain regions. K. Szigeti.

386T Immunotherapy development for *C9ORF72* ALS/FTD using a BAC transgenic mouse model and human antibodies targeting RAN proteins. L. Nguyen.

389T Gene discoveries in autism are biased towards intellectual disability. M. Jensen.

392T Clinical features in a pediatric population due to chromosome deletions at a third level pediatric Mexican hospital in 24 years period of time: Five case reports. M.D. Hurtado-Hernandez.

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398T Using a combined approach of comparative genomic analysis, luciferase assay and CRISPR/Cas9 to identify and characterize cis-regulatory elements in the critical region of the 9p deletion syndrome. X. Hauge.

401T The commitment complex *in vivo* over long intron genes. Y. Leader.

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407T Breakpoint analysis of chromosomes having inverted duplication with terminal deletion by NGS. H. Inagaki.

410T Method for deciding detection limit of droplet digital PCR for rare mutations. T. Takahashi.

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416T Detection of non-reference L1 insertions in the human genome by using a target enrichment method. W. Shin.

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437T Human germline mutation hotspots are characterized by a transversion-rich mutation signature. J. Carlson.

Prenatal, Perinatal, and Reproductive Genetics

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Genetic Counseling, ELSI, Education, and Health Services Research

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Cancer Genetics

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713T ‡ Functional role of intragenic methylation in alternative splicing in cancer. Y. Lee.

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785T Functional integration of genomic and transcriptomic data using Strand NGS explains drug resistance in basal cell carcinoma. S. Kapoor.

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Mendelian Phenotypes

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914T Case report of a mild skeletal phenotype secondary to mutations in *LBR* gene. M.D.F. Carvalho.

917T Spinal manifestations of patients with musculocontractural Ehlers-Danlos syndrome caused by CHST14/D4ST1 deficiency (mcEDS-*CHST14*). T. Kosho.

920T Investigation of the molecular basis of familial and isolated Tarlov cysts. M. Muriello.

923T Early oro-dental manifestations as a clue for the clinical diagnosis of infantile systemic hyalinosis. I.S.M. Sayed.

926T Delineation of musculocontractural Ehlers-Danlos syndrome caused by dermatan sulfate epimerase deficiency (mcEDS-*DSE*): Report of additional patients and comprehensive review of reported cases. A. Unzaki.

929T Cutaneous neurofibromas in neurofibromatosis type 1: A quantitative natural history study. A. Cannon.

932T Synonymous variant in *KCTD7* causes alternative splicing in siblings with progressive epilepsy. D.B. Zastrow.

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938T A novel *PRRT2* pathogenic variant in a family with paroxysmal kinesigenic dyskinesia and benign familial infantile seizures. J.G. Lu.

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1061T Intellectual disability and macrocephaly in three sisters from a consanguineous marriage with a novel *SZT2* homozygous mutation. C. Trujillo.

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1091T *SLC6A6*: Nutritional therapeutic potential of a novel autosomal recessive gene for progressive retinal degeneration and cardiomyopathy. E. Ranza.

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