Concurrent Sessions C13 - C17 - Monday, June 8 - 13.15 - 14.45 hrs

Room	Clyde Auditorium	Hall 5	Hall 2	Lomond Auditorium	Hall 1	Forth Room
	C13. Fundamental insights in structural genomics	**C14. Challenges in genetic counselling**	**C15. Network and functional analysis in intellectual disability**	**C16. Growth failure and microcephaly**	**C17. Epigenetic control of gene expression**	**C18. Metabolic and renal disorders**
13.15	**C13.1** **Human-specific gene evolution and diversity of the chromosome 16p11.2 autism CNV** ***Giuliana Giannuzzi***, *X. Nuttle, M.H. Duyzend, P.H. Sudmant, O. Penn, G. Chiatante, M. Malig, J. Huddleston, L. Denman, L. Harshman, J. Chrast, C. Baker, A. Raja, K. Penewit, F. Antonacci, A. Reymond, E.E. Eichler;* *Lausanne, Switzerland*	**C14.1** **External Quality Assessment of Genetic Counselling: experiences with the first pilot assessment** ***Conny M.A. van Ravenswaaij-Arts***, *C. van Asperen, E. Dequeker, L. Tranebjaerg, L. Garavelli, B. Peterlin, B. Cope, H. Skirton, R. Hastings, ESHG Genetic Services Quality Committee;* *Groningen, Netherlands*	**C15.1** **Genome-wide association study of 200,000 individuals identifies 18 genome-wide significant loci and provides biological insight into human cognitive function** ***Tonu Esko****, *on the behalf of Social Science Genetic Association Consortium (SSGAC);* *Tartu, Estonia*	**C16.1** **Systematic evaluation of patients with idiopathic short stature using whole exome sequencing** ***Christian T. Thiel***, *N.N. Hauer, S. Schuhmann, E. Schöller, M.T. Wittmann, S. Uebe, A.B. Ekici, H. Sticht, H. Dörr, A. Reis;* *Erlangen, Germany*	**C17.1** **RNF12 is essential for X-inactivation in female mouse embryonic stem cells, is required for female mouse development, and might be a target for future therapies to treat X-linked disorders in females: evidence from a mouse knockout model** ***Tahsin Stefan S. Barakat****, *J. Gribnau;* *Rotterdam, Netherlands*	**C18.1** **Disassembly of MINOS complex by *CHCHD10* mutations promotes loss of mitochondrial cristae with defects in mitochondrial genome maintenance and apoptosis** *E. Genin, M. Plutino, S. Bannwarth, E. Villa, E. Cisneros-Barroso, M. Roy, B. Ortega-Vila, K. Fragaki, F. Lespinasse, E. Pinero-Martos, G. Augé, D. Moore, F. Burté, S. Lacas-Gervais, Y. Kageyama, P. Yu-Wai-Man, H. Sesaki, J. Ricci, C. Vives-Bauza,* ***Véronique Paquis-Flucklinger****;* *Nice, France*
13.30	**C13.2** **The impact and activity of mobile elements within the genome** ***Jayne Y. Hehir-Kwa***, *D. Thung, V. Guryev, W.P. Kloosterman, T. Marschall, K. Ye, J.A. Veltman;* *Nijmegen, Netherlands*	**C14.2** **Hereditary breast and ovarian cancer syndrome: successful, large-scale implementation of a group-based approach to genetic counseling.** ***Patrick R. Benusiglio***, *M. Di Maria, A. Jouinot, B. Claret, D. Boinon, D. Lejri, O. Caron;* *Villejuif, France*	**C15.2** **Systematic phenotype-based deconvolution of intellectual disability disorders into biologically coherent modules** ***Christiane Zweier***, *K. Kochinke, B. Nijhof, M. Fenckova, P. Cizek, F. Honti, S. Keerthikumar, M.A.W. Oortveld, T. Kleefstra, J.M. Kramer, C. Webber, M.A. Huynen, A. Schenck;* *Erlangen, Germany*	**C16.2** **Mutations in the core NHEJ components LIG4 and XRCC4 result in microcephalic primordial dwarfism** ***Jennie E. Murray****, *M. van der Burg, H. Ijspeert, P. Carroll, Q. Wu, T. Ochi, A. Leitch, E.S. Miller, B. Kysela, A. Jawad, A. Bottani, F. Brancati, M. Cappa, V. Cormier-Daire, C. Deshpande, E. Ali Faqeih, G. Graham, E. Ranza, T.L. Blundell, A.P. Jackson, G.S. Stewart, L.S. Bicknell;* *Edinburgh, United Kingdom*	**C17.2** **Pattern of X chromosome inactivation across human tissues - insights from population-scale and single-cell RNA sequencing** ***Taru Tukiainen****, *A. Villani, A. Kirby, D. DeLuca, R. Satija, A. Byrnes, J. Maller, T. Lappalainen, The GTEx Project Consortium, A. Regev, K. Ardlie, D. MacArthur;* *Boston, United States*	**C18.2** **COQ4 mutations cause a broad spectrum of mitochondrial disorders associated with CoQ10 deficiency** ***Laura Kremer****, *G. Brea-Calvo, T.B. Haack, D. Karall, A. Ohtake, F. Invernizzi, R. Carrozzo, S. Dusi, C. Fauth, S. Scholl-Bürgi, E. Graf, U. Ahting, N. Resta, N. Laforgia, D. Martinelli, D. Verrigni, Y. Okazaki, M. Kohda, P. Freisinger, T. Strom, T. Meitinger, C. Lamperti, A. Lacson, P. Navas, J. Mayr, E. Bertini, K. Murayama, M. Zeviani, D. Ghezzi, H. Prokisch;* *Neuherberg, Germany*
13.45	**C13.3** **Chromosomal contacts connect loci associated with autism, BMI and head circumference phenotypes** *Maria Nicla Loviglio, M. Leleu, G. Giannuzzi, K. Mannik, E.*	**C14.3** **Experiences of systematic genetic testing involving women recently diagnosed with epithelial ovarian cancer: a qualitative study** ***Hannah E. Shipman***,	**C15.3** **9.6% of mouse gene knockouts show abnormal neuroanatomy: a resource to identify genes and gene networks involved in ID in human** *B. Yalcin,* ***Anna Mikhaleva****, V.E.*	**C16.3** **Loss-of-Function Mutations in WDR73 Are Responsible for Microcephaly and Steroid-Resistant Nephrotic Syndrome: Galloway-Mowat Syndrome** ***Estelle Colin****, *E. Huynh Cong, G. Mollet, A. Guichet, O. Gribouval, C. Arrondel, O. Boyer, L. Daniel, M. Gubler, Z. Ekinci, M. Tsimaratos, B. Chabrol, N. Boddaert, A. Verloes, A. Chevrollier, N. Gueguen, V. Desquiret-Dumas, M. Ferré, V. Procaccio, L. Richard, B. Funalot, A. Moncla, D.*	**C17.3** **Genome wide DNA promoter methylation: Differences in human subcutaneous vs. omental visceral adipose tissue** ***Maria Keller****, *L. Hopp, X. Liu, K. Rohde, M. Klös, A. Dietrich, M. Schön, D. Gärtner, T. Lohmann, M. Dreßler, M. Stumvoll, P. Kovacs, H. Binder, M. Blüher, Y. Böttcher;*	**C18.3** **MCT1 deficiency impairs ketone utilization and causes profound ketoacidosis upon catabolic stress** *P. van Hasselt, S. Ferdinandusse, G. Monroe, J. Ruiter, M. Turkenburg, M. Geerlings, K. Duran, M. Harakalova, B. van der Zwaag, A. Monavari, I. Okur, M. Sharrard, M. Cleary, N. O'Connell, V. Walker, E. Rubio*

	*Migliavacca, I. Roberts-Caldeira, I. van der Werf, 16p11.2 European Consortium, J.S. Beckmann, S. Jacquemont, J. Rougemont, **A. Reymond**; Lausanne, Switzerland*	*M. Tischkowitz, S. Flynn, C. MacDonald-Smith, N. Hulbert-Williams, GTEOC Study team; Cambridge, United Kingdom*	*Vancollie, M. Kannan, H. Whitley, A. Edwards, C. Wagner, J. Estabel, C.J. Lelliott, J.K. White, Sanger Mouse Genetics Project, D.J. Adams, D.A. Keays, J. Flint, Y. Herault, A. Reymond; Lausanne, Switzerland*	*Bonneau, C. Antignac; Angers, France*	*Leipzig, Germany*	*Gozalbo, M. de Vries, G. Visser, R. Houwen, J. van der Smagt, N. Verhoeven-Duif, R. Wanders, **Gijs van Haaften**; Utrecht, Netherlands*
14.00	**C13.4** **Single-cell allele specific expression (ASE) in T21: a novel approach to understand Down syndrome.** ***Georgios Stamoulis**, P. Makrythanasis, F. Santoni, A. Letourneau, M. Guipponi, M. Garieri, N. Panousis, E. Falconnet, P. Ribaux, C. Borel, S.E. Antonarakis; Geneva, Switzerland*	**C14.4** **Sharing information with children and young people about adult-onset inherited conditions: Using evidence to improve services for parents and their children** ***Karen Forrest Keenan**, L. McKee, Z. Miedzybrodzka; Aberdeen, United Kingdom*	**C15.4** **Finding new connections in the transcriptional regulation of Lysine-specific demethylase 5C (KDM5C) a disease gene involved in syndromic and non-syndromic XLID** ***Agnese Padula**, L. Poeta, C. Shoubridge, A. Ranieri, K. Helin, J. Gecz, C. Schwartz, M.V. Ursini, H. vanBokhoven, M.G. Miano; Naples, Italy*	**C16.4** **Mutations in PLK4, encoding a master regulator of centriole biogenesis, and its substrate, TUBGCP6, cause microcephaly, growth failure and retinopathy** ***Louise S. Bicknell**, C. Martin, A. Klingseisen, I. Ahmad, M.S. Hussain, A. Leitch, G. Nurnberg, M.R. Toliat, J. Murray, D. Hunt, F. Khan, Z. Ali, S. Tinschert, J. Ding, C. Keith, M.E. Harley, P. Heyn, R. Mueller, I. Hoffman, V. Cormier-Daire, H. Dollfus, L. Dupuis, A. Bashamboo, K. McElreavey, A. Kariminejad, R. Mendoza-Londono, A.T. Moore, A. Saggar, C. Schlechter, R. Weleber, H. Thiele, J. Altmuller, W. Hohne, M.E. Hurles, A.A. Noegel, S.M. Baig, P. Nurnberg, A.P. Jackson; Edinburgh, United Kingdom*	**C17.4** **Mapping genetic and epigenetic factors influencing human hippocampal gene expression** ***Andrea Hofmann**, H. Schulz, A. Ruppert, S. Herms, K. Pernhorst, C. Wolf, N. Kerbalai, O. Stegle, D. Czamara, S. Sivalingam, A. Hillmer, B. Pütz, A. Woitecki, S. Schoch, A.J. Forstner, B. Müller-Myhsok, M.M. Nöthen, T. Sander, A. Becker, P. Hoffmann, S. Cichon; Bonn, Germany*	**C18.4** **Rare non-synonymous variations in the human ferroportin iron transporter gene (haemochromatosis type 4): the quest for causal mutations** *I. Callebaut, S. Pissard, C. Kannengiesser, V. Gérolami, C. Ged, F. Cartault, J. Rochette, C. Ka, C. Férec, **Gérald Le Gac**; Brest, France*
14.15	**C13.5** **High incidence of mosaic chromosomal aneuploidies in human cell lines: a quantification of the frequency of the phenomenon** ***Eftychia S. Dimitriadou**, M. Zamani Esteki, N. Van der Aa, T. Voet, J.R. Vermeesch; Leuven, Belgium*	**C14.5** **Attitudes towards returning data to participants in sequencing research** ***Anne Middleton**, C. Wright, H. Firth, M. Hurles, M. Parker, on behalf of the DDD study; Cambridge, United Kingdom*	**C15.5** **HCFC1 is a dosage sensitive transcriptional coregulator of neurodevelopment that influences neural progenitor and neuronal cell function** ***Lachlan A. Jolly**, L.S. Nguyen, D. Domingo, Y. Sun, S. Barry, M. Hancarova, P. Plevova, M. Vlckova, M. Havlovicova, V.M. Kalscheuer, C. Graziano, T. Pippucci, Z. Sedlacek, E. Bonora, J. Gecz; Adelaide, Australia*	**C16.5** **Mutations in TUBGCP4 alter microtubule organization via the ?-tubulin ring complex ?TuRC in autosomal recessive microcephaly with chorioretinopathy.** ***Sophie Scheidecker**, C. Etard, L. Haren, C. Stoetzel, S. Hull, G. Arno, V. Plagnol, S. Drunat, S. Passemard, A. Toutain, C. Obringer, M. Koob, V. Geoffroy, V. Marion, U. Strähle, P. Ostergaard, A. Verloes, A. Merdes, A.T. Moore, H. Dollfus; Strasbourg, France*	**C17.5** **Analysis of monoallelic expression in human individual cells revealed novel imprinting genes.** ***Christelle Borel**, F. Santoni, M. Garieri, E. Falconnet, P. Ribaux, S.E. Antonarakis; Geneva, Switzerland*	**C18.5** **Companion diagnostics by comprehensive targeted NGS with evidence for a threshold model in a cohort of 605 patients with atypical haemolytic uremic syndrome and hereditary glomerulopathies** *M. Grohmann, N. Bachmann, M. Hiersche, T. Eisenberger, H.J. Bolz, T. Ring, B. Hohenstein, C. Mache, M.J. Kemper, C.S. Haas, N. Heyne, R.P. Wüthrich, F. Thaiss, B. Tönshoff, L. Pape, M. Wiesener, J. Menne, G. Walz, **Carsten Bergmann**; Ingelheim, Germany*
14.30	**C13.6** **Chromothripsis in healthy individuals affects multiple protein-coding genes and can result in severe congenital abnormalities in offspring** ***Mirjam S. de Pagter**, M.J. van Roosmalen, A.F. Baas, I. Renkens, K.J. Duran, E. van Binsbergen, M. Tavakoli-Yaraki, R. Hochstenbach, L.T.*	**C14.6** **Population-based Preconception Carrier Screening: how do potential users view a preconception test for 70 severe autosomal recessive diseases?** ***Mirjam Plantinga**, E. Birnie, S. Kaplan, M.A. Verkerk, A.M. Lucassen, A.V.*	**C15.6** **Clinical and experimental evidence establish a link between *KIF7* and *C5orf42*-related ciliopathies** ***Reza Asadollahi**, J.E. Strauss, M. Zenker, O. Beuing, S. Edvardson, O. Elpeleg, P. Joset, T.M. Strom, D. Niedrist, B. Oneda, S. Azzarello-Burri, M. Papik, A. Baumer, K. Steindl, A. Schinzel, E.T. Stoeckli, A. Rauch; Zurich-Schlieren,*	**C16.6** **From whole exome sequencing to functional studies in syndromic microcephaly: using zebrafish for variant testing** ***F. Cristofoli**, E. E. Davis, K. Devriendt, H. Peeters, H. Van Esch, J. R. Vermeesch; Leuven, Belgium*	**C17.6** **Novel method reveals a large number of expression quantitative trait loci (eQTLs) influencing transcript levels in a Parent-of-origin fashion** ***Aaron F. McDaid**, T. Esko, L. Franke, Z. Kutalik; Lausanne, Switzerland*	**C18.6** **Common and rare variants associated with kidney stones and biochemical traits** ***Asmundur Oddsson**, P. Sulem, H. Helgason, V. Edvardsson, G. Thorleifsson, G. Sveinbjornsson, E. Haraldsdottir, G.I. Eyjolfsson, O. Sigurdardottir, I. Olafsson, G. Masson, H. Holm, D.F. Gudbjartsson, U. Thorsteinsdottir, O.S. Indridason, R. Palsson, K. Stefansson; Reykjavik, Iceland*

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City & Country refer to presenting authors

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