Conference Programme

(☆ Denote early career researcher presentations)

Sunday 16th November 2025

12:00 - 13:55 Registration, Lunch, Poster Mounting and E-posters go live

13:55 - 14:00 Welcome and Conference Opening

14:00 - 15:30 Oral Presentations A - **Lumping, Splitting and Allelic Disorders**

Chairs - Fowzan Alkuraya, Dian Donnai

14:00 - 14:30 (Oral 01) Lumping and Splitting in the Molecular Age
Leslie Biesecker

14:30 - 14:45 (Oral 02) Deleterious *ZNRF3* germline variants as a novel cause of neurodevelopmental disorders with mirror brain phenotypes due to distinct domain-specific effects on Wnt/β-catenin signaling
Anita Rauch

14:45 - 15:00 (Oral 03) *PLCB4* dominant negative, loss-of-function and gain-of-function variants leading to auriculocondylar syndrome or a novel syndrome
Jeanne Amiel

15:00 - 15:10 (Oral 04) Houge-Janssens syndrome types 1-4: Clinical overview and novel pathogenic mechanisms
Gunnar Douzgos Houge

15:10 - 15:20 (Oral 05) Delineating *TAOK* Gene Disorders: Distinct Neurodevelopmental disorders associated with *TAOK1*, *TAOK2*, and *TAOK3* Variants
☆Nour Elkhateeb

15:20 - 15:30 (Oral 63) *PUF60*-related disorders, new insights using RNA sequencing suggest biological pathways and disease signature
Diana Baralle

15:30 - 16:15 **Coffee**

16:15 - 17:45 Oral Presentations B - **Environment, Exposures and Imprinting**

Chairs - Kate Baker, Sofia Douzgou Houge

16:15 - 16:45 (Oral 07) The Fetal Fentanyl syndrome - delineation of the phenotype in 37 infants prenatally exposed to fentanyl
Miguel del Campo

16:45 - 17:00 (Oral 08) Discovery of a DNA methylation episignature as a diagnostic biomarker for Fetal Alcohol Syndrome
Mieke van der Haelst

17:00 - 17:15 (Oral 09) Investigating the molecular basis of multi-locus imprinting disturbance
Eamonn Maher

17:15 - 17:25 (Oral 10) Understanding the impact of MLID on imprinting disorders: preliminary results from the StratifID study
Gabriella Gazdagh

17:25 - 17:35 (Oral 11) Neuropsychological Functioning in Fetal Valproate Spectrum Disorder
Rebecca Bromley

17:35 - 17:45 (Oral 12) Infantile Caffey's Disease a genetic mimic. Another disease caused by transplacental transfer of IgG antibodies. MGUS with significance
Tessa Homfray

19:00 - 22:00 **Cloud 23 Reception and Buffet Dinner**

Monday 17th November 2025

09:00 - 10:30 Oral Presentations C - **Epigenomics of Rare Diseases (EpiGenRare Symposium)**

Chairs - Siddharth Banka, Cristina Dias

09:00 - 09:30 (Oral 13) A novel segmental ageing syndrome demonstrates DNA methylation causes multiple age-related pathologies
Andrew Jackson

09:30 - 09:45 (Oral 14) A novel neurodevelopmental syndrome caused by loss-of-function of the *DMAP1* gene
Dong Li

09:45 - 10:00 (Oral 15) Novel Mendelian Neurodevelopmental syndrome caused by *de novo* variants in *ATAD2B* phenotypically recapitulated in *Atad2b*$^{-/-}$ mice
Elizabeth Bhoj

10:00 - 10:10 (Oral 16) Deficiency of the histone lysine demethylase *KDM5B* alters histone methylation and gene expression in the developing brain and causes autism-like phenotypes via increased NMDAR signalling
Albert Basson

10:10 - 10:20 (Oral 17) *FBXO22* deficiency defines a pleiotropic syndrome of growth restriction and multi-system anomalies associated with a unique epigenetic signature
Aisha Al Shamsi

10:20 - 10:30 (Oral 18) Improving diagnosis and mechanistic understanding of *KMT2D*-related branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism BCAHH syndrome
Sara Cuvertino

Rare Disease Research UK.
Epigenomics of Rare Disorders - EpiGenRare

Join the EpiGenRare network by scanning the QR code

10:30 - 11:15 **Coffee**

11:15 - 12:45 Oral Presentations D - **The Non-coding, Regulatory and Genomic Disorders**

Chairs - Stefan Barakat, Jamie Ellingford

11:15 - 11:30 (Oral 19) Analysis of R-loop forming regions discovers *RNU2-2* and *RNU5B-1* as non-coding neurodevelopmental disorder genes
☆Adam Jackson

11:30 - 11:45 (Oral 20) Biallelic variants in the non-coding RNA gene *RNU4-2* cause a recessive neurodevelopmental syndrome with distinct white matter changes
☆Alexander Blakes

11:45 - 12:00 (Oral 21) Rare variants disrupting mRNA cleavage and polyadenylation associated with undiagnosed rare disorders
Yaroslav Kainov

12:00 - 12:15 (Oral 22) Exploring TAD-related *FGF* gene dysregulation by structural/copy number variants in craniosynostosis using differentiated iPSCs, RNA-seq, ATAC-seq and mouse models
Andrew Wilkie

12:15 - 12:30 (Oral 23) Synthetic regulatory landscapes in vivo for uncovering effects of noncoding mutations in congenital eye disease
Shipra Bhatia

12:30 - 12:45 (Oral 24) Duplication-triplications on 16p13.3 cause a recognisable neurodegenerative disorder with ataxia: the second genomic disorder linked to a 144kb palindrome.
☆James Fasham

12:45 - 14:00 **Lunch**

14:00 - 15:30 **Poster Session A**

15:30 - 17:00 Oral Presentations E - **Metabolic and Endocrine Disorders**

Chairs - Tiong Yang Tan, Charulata Deshpande

15:30 - 15:45 (Oral 25) Bi-allelic *FKBP4* loss of function variants cause a syndrome of sexual differentiation disorder and global developmental delay
☆John McDermott

15:45 - 16:00 (Oral 26) A novel and severe multisystem metabolic disease due to homozygous *AIP* variants
Hilde Van Esch

16:00 - 16:15 (Oral 27) Early initiation of sulphonylurea therapy improves neurodevelopmental outcomes in individuals with *KCNJ11*-related iDEND syndrome (developmental delay, epilepsy and neonatal diabetes)
☆Pamela Bowman

16:15 - 16:30 (Oral 28) Biallelic variants in *LSR* cause a microcephalic neurodevelopmental disorder with cholestatic liver disease
☆Joseph Leslie

16:30 - 16:45 (Oral 29) Bi-allelic *UGGT1* variants cause a congenital disorder of glycosylation
Emma L Baple

16:45 - 17:00 (Oral 30) *De novo* mutations in *XRN1* cause a novel dominant form of lethal mitochondrial cardiomyopathy
Robert Taylor

Tuesday 18th November 2025

09:00 - 10:30 Oral Presentations F - **Vascular and Connective Tissue**

Chairs - Bert Callewaert, Emma Burkitt Wright

09:00 - 09:15 (Oral 32) Monoallelic *FNDC3B* variants cause a novel connective tissue disorder with aortopathy and other vascular tortuosity
Oana Caluseriu

09:15 - 09:30 (Oral 33) An integrative machine learning approach to classify cutis laxa patients, supported by electron microscopy
Bert Callewaert

09:30 - 09:45 (Oral 34) Safety findings from the phase 1/2 MOSAIC study of miransertib for patients with *PIK3CA*-related overgrowth spectrum or Proteus syndrome
Himanshu Goel

09:45 - 10:00 (Oral 35) Changing landscape of clinical dysmorphology using model systems for advancing diagnostics and therapeutics in rare connective tissue disorders
Meena Balasubramanian

10:00 - 10:30 (Oral 31) New therapeutic approaches for KRAS-related vascular malformations
Guillaume Canaud

10:30 - 11:15 **Coffee**

11:15 - 12:45 Oral Presentations G - **Developmental Epileptic Disorders**

Chairs - Elizabeth Bhoj, Abhijit Dixit

11:15 - 11:30 (Oral 36) *SETD1B* variants drive neurodevelopmental disorders and epilepsy: delineation of the clinical phenotype of more than 120 patients and insights from 2D and 3D human models
Stefan Barakat

11:30 - 11:45 (Oral 37) All pediatric patients with Bachmann-Bupp syndrome treated with repurposed eflornithine demonstrate clinical and biochemical improvement
☆Elizabeth VanSickle

11:45 - 11:55 (Oral 38) L-Serine treatment in patients with *GRIN* loss-of-function variants, experience from a non academic genetic centre
Damien Lederer

11:55 - 12:05 (Oral 39) Hydroxyglutaric aciduria type II in an infant: One year experience with experimental treatment with enasidenib
Sofia Douzgou Houge

12:05 - 12:15 (Oral 40) Homozygous *MDGA2* loss-of-function variants cause developmental and epileptic encephalopathy
Heba Morsy

12:15 - 12:25 (Oral 41) Biallelic *WDR91* variants cause a severe early-onset neurodevelopmental disorder with refractory epilepsy
☆Giulia Spoto

12:25 - 12:35 (Oral 06) A decade-long journey to uncover a novel mechanism: when the well-known *TCF4* gene tells a different story
Laurence Faivre

12:35 - 12:45 (Oral 43) Identification of *TCP1* variant and variants in other components of the TRiC chaperonin complex associated with brain malformations and seizures
Marije Meuwissen

12:45 - 14:00 **Lunch**

14:00 - 15:30 **Poster Session B**

15:30 - 17:00 Oral Presentations H - **Phenotyping and Cohort Studies**

Chairs - A Micheil Innes, Gijs Santen

15:30 - 15:40 (Oral 44) *GNAI2* mutations in humans: clinical presentations and immune dysregulation in a cohort of 20 patients
Helen Stewart

15:40 - 15:50 (Oral 45) An International *ASXL3* Quasi-Natural History Study; Largest Cohort, Novel Associations and Clinical Recommendations
☆Emily Woods

15:50 - 16:00 (Oral 46) Missense variants in *ARID1A* and *ARID1B*: Location, location, location?
Gijs Santen

16:00 - 16:10 (Oral 47) CHID (Choroid plexus hyperplasia, Hydrocephalus and Intellectual Disability) patients share a distinct clinical phenotype and methylation episignature
☆Fiona Stirling

16:10 - 16:20 (Oral 48) Brain MRI dysmorphology in uncovering tubulinopathies -multigenerational inheritance of *TUBB*-related tubulinopathy
Abhijit Dixit

16:20 - 16:30 (Oral 49) Expanding the genetic and phenotypic spectrum of *SAMD9* and MIRAGE syndrome: a UK cohort study
Emma Wakeling

16:30 - 16:40 (Oral 50) *ELFN1* Deficiency: the mechanistic basis and phenotypic spectrum of a neurodevelopmental disorder with epilepsy
☆Rhys Dore

16:40 - 16:50 (Oral 51) Phenotypic spectrum of biallelic *DIAPH1* deficiency.
Mohnish Suri

16:50 - 17:00 (Oral 52) The common A391T hypomorphic variant in *SLC39A8* is a significant contributor to the architecture of autosomal recessive (AR) *SLC39A8*-CDG (CDG2N)-a recurrent treatable neurodevelopmental disorder (NDD) associated with manganese deficiency
A Micheil Innes

19:00 - 23:00 **Conference Dinner and Party**

Wednesday 19th November 2025

09:00 - 10:30 Oral Presentations I - **Large Scale and Global Genomics**

Chairs - Leslie Biesecker, Bill Newman

09:00 - 09:30 (Oral 53) Adult genomic medicine: lessons from a multisite study of 2,700 patients
Fowzan Alkuraya

09:30 - 09:45 (Oral 54) PhenomAD-NDD: revealing comorbidities in 51,227 children with neurodevelopmental disorders
Bert de Vries

09:45 - 10:00 (Oral 55) Assessment of uniparental disomy in whole genome sequence datasets for 46,955 families implicates a significant role in rare disease aetiology
Jamie Ellingford

10:00 - 10:10 (Oral 56) Identification of Human Knockouts in 34 Candidate Genes Potentially Linked to Severe Neurodevelopmental Disorders
Ambrin Fatima

10:10 - 10:20 (Oral 57) Deciphering Developmental Disorders in Africa (DDD-Africa) Study - successes and developments in an African Setting
Amanda Krause

10:20 - 10:30 (Oral 58) DDD-Africa: the Congolese experience of introducing Exome Sequencing as a first-tier diagnostic test for unexplained Developmental Disorders
☆Prince Makay, Koenraad Devriendt

10:30 - 11:15 **Coffee**

11:15 - 12:45 Oral Presentations J - **Neurodevelopmental Syndromes**

Chairs - Bert de Vries, Emma Baple

11:15 - 11:30 (Oral 59) Biallelic variants in *GTF3C1* encoding RNA polymerase transcription factor III in individuals with neurodevelopmental disability, microcephaly, cerebellar hypoplasia and craniofacial anomalies
Tiong Yang Tan

11:30 - 11:45 (Oral 60) Deciphering how the cellular phenotype associated with biallelic pathogenic variants in *SMC5* and *SMC6* affects clinical presentation
☆Charlotte Sherlaw-Sturrock

11:45 - 11:55 (Oral 61) Expanding the Clinical and Molecular Landscape of BAF Complex Intellectual Developmental Disorders
Cristina Dias

11:55 - 12:05 (Oral 62) Emotion regulation in children with rare ID-associated genetic conditions: a multi-method approach
Kate Baker

12:05 - 12:15 (Oral 42) Variants in *LRRC4* are associated with predominant speech delay, intellectual disability and alterations in neuronal morphology and synapse numbers.
☆Lauren Cairns

12:15 - 12:25 (Oral 64) *CAPN8* is associated with a novel syndromic congenital muscular dystrophy
☆Lettie Rawlins

12:25 - 12:35 (Oral 65) *ZFHX4* loss-of-function variants define a recognisable neurodevelopmental syndrome with consistent facial dysmorphism and multisystem involvement
Fernando Santos-Simarro

12:35 - 12:45 (Oral 66) Biallelic variants in *JKAMP* cause a novel neurodevelopmental disorder with urogenital anomalies
Reham Khalaf-Nazzal

12:45 - 14:00 **Lunch**

14:00 - 15:30 Oral Presentations K - **Skeletal Disorders**

Chairs - Mohnish Suri, Kay Metcalfe

14:00 - 14:15 (Oral 70) A novel form of autosomal dominant spondylocostal dysostosis in three unrelated families caused by the same heterozygous pathogenic variant in *MESP2*
Geert Mortier

14:15 - 14:30 (Oral 69) Modelling human growth plate cartilage using Pluripotent Stem Cells
Steven Woods

14:30 - 14:45 (Oral 68) The absence of an enzyme-rescue metabolite as the cause of Catel-Manzke syndrome
Nadja Ehmke

14:45 - 15:00 (Oral 71) Efficacy of TXAS inhibitors to improve bone mineral density and quality in Osteogenesis imperfecta
Valérie Cormier-Daire

15:00 - 15:30 (Oral 67) New precision treatments for children with achondroplasia: a template for rare disease therapies
Ravi Savarirayan

15:30 - 15:45 Prizes and Conference Close

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