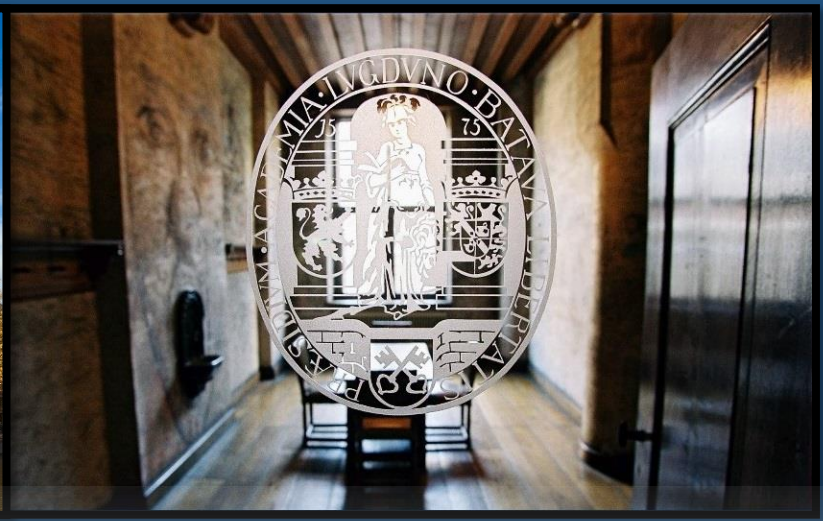
PROGRAMME

3rd International Workshop on Klinefelter Syndrome, Trisomy X and XYY

12-14 September 2022, Leiden, The Netherlands



Programme themes

Genetics and X chromosome dynamics

Endocrinology and fertility

Developmental impact in childhood and adolescence

Impact of X and Y on life course

Future perspectives, treatment and guidelines

Social events

Welcome dinner at the National Museum of Antiquities Leiden

Boat tour in the historic city center of Leiden

Farewell lunch at the historic botanical gardens of the Academy Building

Universiteit Leiden
The Netherlands

TRIXY
Center of Expertise

European City of Science Leiden2022

DAY 1 – Monday September 12

Venue: Academy Building (Telders Auditorium), Rapenburg 73, Leiden

Registration and opening			
9.00-9.30	Registration		
9.30-10.00	Opening	Hanna Swaab	
Genetics and X chromosome dynamics			**Chair: Claus Gravholt**
10.00-10.45	Keynote presentation O1	Anne Skakkebæk	The genetic architecture of sex chromosome aneuploidies – new developments
10.45-11.30	Keynote presentation O2	Armin Raznahan	Sex Chromosome Dosage Effects: From the Genome to the Brain
11.30-12.00	*Short break*		
12.00-12.20	Oral presentation O3	Emma Johannsen	Sex chromosome aneuploidies give rise to pervasive changes in the circular RNA profile: A circular transcriptome-wide study of Turner and Klinefelter syndrome across different tissues
12.20-12.40	Oral presentation O4	Marco Barchi	The proper interplay between the expression of Spo11 splice isoforms and the structure of the pseudoautosomal region leads XY recombination
12.40-13.00	Oral presentation O5	Antonio Adamo	The impact of X-polysomy during early embryogenesis in Klinefelter Syndrome and High-grade X chromosome aneuploidy
13.00-14.00	*Posters & lunch*		
Endocrinology and fertility			**Chair: Jörg Gromoll**
14.00-14.45	Keynote presentation O6	Alberto Ferlin	Testis function, hypogonadism and bone metabolism
14.45-15.30	Keynote presentation O7	Anders Juul	Klinefelter in transition: auxological, endocrine and metabolic changes during puberty
15.30-16.00	*Short break*		
16.00-16.20	Oral presentation O8	Martin den Heijer	Aging in Klinefelter patients
16.20-16.40	Oral presentation O9	Sofia Boeg Winge	Focal spermatogenesis in men with Klinefelter syndrome requires loss of XIST expression and of the additional X-chromosome in Sertoli cells
16.40-17.00	Oral presentation O10	Shanlee Davis	Randomized-controlled trial of testosterone in 70 infants with 47,XXY
17.00-17.15	*Short break*		
Fertility			**Chair: Alberto Ferlin**
17.15-18.00	Keynote presentation O11	Alan Rogol	Sex Chromosome Aneuploidies and Fertility: 47 XXY, 47XYY, 47XXX, and 45X/47XXX
Social event			
19.00	*Welcome dinner at the National Museum of Antiquities Leiden (Rapenburg 28, Leiden)*		

National Museum of Antiquities, Rapenburg 28, Leiden

DAY 2 – Tuesday September 13

Venue: Academy Building (Telders Auditorium), Rapenburg 73, Leiden

Opening			**Chair: Hanna Swaab and Sabine Hannema**
8.30-9.00	Walk-in		
9.00-10.00	Oral presentation	Experience experts	Living with an extra X or Y
Developmental impact in childhood and adolescence			**Chair: Alberto Ferlin**
10.00-10.45	Keynote presentation O12	Sophie van Rijn	Neurocognitive and behavioral development in young children (1-7 years) with Sex Chromosome Trisomy: the TRIXY Early Childhood Study
10.45-11.30	Keynote presentation O13	Nicole Tartaglia	The eXtraordinarY Babies Study: A Prospective Natural History Study of Health and Neurodevelopment in Children with a Prenatal Diagnosis of Sex Chromosome Trisomy
11.30-12.00	*Short break*		
12.00-12.20	Oral presentation O14	Shanlee Davis	System-Based Diagnoses in Youth with XXY, XXY, and XXX
12.20-12.40	Oral presentation O15	Laura Zampini	Oral narrative skills in preschool children with sex chromosome trisomies
12.40-13.00	Oral presentation O16	Kaat Theelen	Neuropsychiatric difficulties in Klinefelter Syndrome and supporting tool checklist
13.00-14.00	*Posters & lunch*		
Impact of X and Y on life course			**Chair: Anders Juul**
14.00-14.45	Keynote presentation O17	Claus Gravholt	Morbidity of 47,XXY and 47,XYY syndromes - similarities and differences
14.45-15.30	*Posters*		
15.30-16.00	*Short break*		
16.00-16.20	Oral presentation O18	Susan Howell	Non-Invasive Prenatal Testing (NIPT) Results for Participants of the eXtraordinarY Babies Study: Screening, Counseling, Diagnosis, and Discordance
16.20-16.40	Oral presentation O19	Francesco Carlomagno	The onset and progression of testicular dysfunction in Klinefelter Syndrome
16.40-17.00	Oral presentation O20	Caroline Harrison	Psychological Effects of Testosterone in Tanner 2-3 Males with XXY: Results of a randomized, placebo-controlled trial
17.00-17.15	*Short break*		
Future perspectives			**Chair: Martin den Heijer**
17.15-18.00	Keynote presentation O21	Jörg Gromoll	Klinefelter Syndrome- a novel view into testicular function
Social event			
18.30	*Boat tour in the historic city center of Leiden including snacks & drinks (starts at Academy Building)*		

DAY 3 – Wednesday September 14

Venue: Academy Building (Hortus Botanicus), Rapenburg 73, Leiden

Treatment & guidelines			**Chair: Nicole Tartaglia**
9.30-10.00	Walk-in		
10.00-10.45	Oral presentation	Alberto Ferlin	Presentation of the 2021 EAA guideline
10.45-11.05	Oral presentation	Lise Aksglæde	Areas that need more focus in Klinefelter syndrome
11.05-11.45	Oral presentation	Shanlee Davis	arguments for new international comprehensive guidelines
11.45-12.15	Oral presentation	Claus Gravholt	Roadmap to new guidelines with proposal for subcommittees
Social event			
12.30-13.30	*Farewell lunch at the historic botanical gardens of the Academy Building*		

Poster Presentations

Abstract #	Presenting author	Title
POSTER PRESENTATIONS		
P1	Charlotte Jakes	Gender identity in Klinefelter Syndrome
P2	Mohamed Al-Hussini	Hormone stimulation treatment for infertility in Klinefelter Syndrome - patient information
P3	Mohamed Al-Hussini	Psychosexual issue affecting men with Klinefelter Syndrome seeking fertility management
P4	Ramzy Elnabarawy	Risk of thromboembolism, and testosterone therapy in KS patients
P5	Shanlee Davis	Unique plasma metabolite signature for adolescents with Klinefelter syndrome reveals altered fatty acid metabolism
P6	Shanlee Davis	Health risks among youth with SCT in a large cohort
P7	Shanlee Davis	Evidence of Leydig Cell Dysfunction in Infants with 47,XXY During the Mini-Puberty of Infancy
P8	Shanlee Davis	Testosterone treatment during the mini-puberty period of infancy affects the hypothalamic-pituitary-gonadal axis
P9	Shanlee Davis	Males with a 47,XXY and 47,XYY genotype who are not clinically identified still have significant medical comorbidities
P10	Susan Howell	Eosinophilic esophagitis in individuals with sex chromosome aneuploidies: Clinical presentations and management implications
P11	Joachim Wistuba	Testicular architecture of patients with SCO caused by 46,XX testicular DSD, Klinefelter syndrome and AZF deletions
P12	Chaira Serrarens	Resting-state functional connectivity in adult women with 47,XXX: a 7 Tesla MRI study
P13	Margo Willems	Identifying KS-specific fibrotic genes by analyzing transcriptome of (non-)fibrotic testicular tissue
P14	Alex Murray	Description of Klinefelter sample
P15	Francesco Carlomagno	The dose-effect of supernumerary X chromosomes on clinical, metabolic and cardiac outcomes of male subjects
P16	Francesco Carlomagno	Altered thyroid feedback loop in Klinefelter syndrome: a longitudinal study from infancy, through transition-age to adulthood
P17	Simon Chang	Increased complement activation in men with Klinefelter syndrome
P18	Jörg Gromoll	Supervised machine learning based prediction models for identification of Klinefelter Syndrome
P19	Jesper Just	Adopting machine learning techniques to describe the phenotype of Klinefelter Syndrome using multiple categories of clinical measurements
P20	Megan Louderman	Temperament of very young children with SCT
P21	Margherita Vecchio	A review of the relationship between Klinefelter Syndrome and Gender Dysphoria
P22	Talia Thompson	Educational Supports for Children With Sex Chromosome Aneuploidies in the U.S.
P23	Kimberly Kuiper	Emotional reactivity and expressivity in young children with sex chromosome trisomies: evidence from psychophysiological and observational data
P24	Sophie van Rijn	The early impact of Sex Chromosome Trisomy (47XXX, 47XXY, 47XYY) on social cognition: evidence from eyetracking
P25	Rebecca Wilson	The eXtraordinarY Babies Study: Early Social Communication Skills in Infants with Sex Chromosome Trisomy (SCT)
P26	Evelien Urbanus	Social communication in children with sex chromosome trisomy (XXY, XXX, XYY): How children attend and respond to short communicative interactions
P27	Jennifer Janusz	The eXtraordinarY Babies Study: Early Adaptive Skills Profile of Infants and Toddlers with Prenatally Identified Sex Chromosome Trisomies
P28	Maarten Otter	Psychiatric symptoms in women with triple X-syndrome (TXS)
P29	Talia Thompson	An International Survey of Academic and Character Strengths in Youth with Sex Chromosome Aneuploidies
P30	Talia Thompson	Parenting young daughters with prenatally identified trisomy X
P31	Nienke Bouw	Early preventive intervention for young children with Sex Chromosome Trisomies (XXX, XXY, XYY)
P32	Francien Martin	Social Management Training for men with Klinefelter Syndrome
P33	Sabrina Cinot	Neuropsychiatric assessment of a post zygotic mosaicism – 45x0/47xyy: a case report
VIDEO PITCHES		
V1	Kate Attfield	Social experience of girls and women with Triple X
V2	Jessica Langenhoff	Peergroups on Facebook: an Xtra valuable option for information and interaction in Trisomy X