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







DAY 1 - 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					
10.00-10.45	Keynote presentation	Ann Skakkebæk	The genetic architecture of sex chromosome aneuploidies – new developments		
10.45-11.30	Keynote presentation	Armin Raznahan	Sex Chromosome Dosage Effects: From the Genome to the Brain		
11.30-12.00	Short break				
12.00-12.20	Oral presentation	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	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	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				
14.00-14.45	Keynote presentation	Alberto Ferlin	Testis function, hypogonadism and bone metabolism		
14.45-15.30	Keynote presentation	Anders Juul	To be announced		
15.30-16.00	Short break				
16.00-16.20	Oral presentation	Martin den Heijer	Aging in Klinefelter patients		
16.20-16.40	Oral presentation	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	Shanlee Davis	Randomized-controlled trial of testosterone in 70 infants with 47,XXY		
17.00-17.15	Short break				
Fertility					
17.15-18.00	Keynote presentation	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 - September 13

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

Opening	Opening					
8.30-9.00	Walk-in					
9.00-10.00	Oral presentation	Experience experts	Living with an extra X or Y			
Developmen	Developmental impact in childhood and adolescence					
10.00-10.45	Keynote presentation	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	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	Shanlee Davis	System-Based Diagnoses in Youth with XXY, XXY, and XXX			
12.20-12.40	Oral presentation	Laura Zampini	Oral narrative skills in preschool children with sex chromosome trisomies			
12.40-13.00	Oral presentation	Stephanie Vanclooster	Neuropsychiatric difficulties in Klinefelter Syndrome and supporting tool checklist			
13.00-14.00	Posters & lunch					
Impact of X a	nd Y on life course					
14.00-14.45	Keynote presentation	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	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	Carlotta Pozza	The onset and progression of testicular dysfunction in Klinefelter Syndrome			
16.40-17.00	Oral presentation	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						
17.15-18.00	Keynote presentation	Jorg Gromoll	Klinefelter Syndrome- a novel view into testicular function			
Social event						
18.30	Boat tour in the historic	Boat tour in the historic city center of Leiden including snacks & drinks (starts at Academy Building)				

DAY 3 - September 14

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

Treatment & guidelines				
9.30-10.00	Walk-in			
10.00-10.45	Keynote presentation	To be announced		
10.45-11.30	Keynote presentation	To be announced		
11.30-12.00	Short break			
12.00-12.30	Concluding discussion			
Social event				
12.30-13.30	Farewell lunch at the historic botanical gardens of the Academy Building			

Poster Presentations

Poster #	Presenting author	Title
P1	Charlotte Jakes	Gender identity in Klinefelter Syndrome
P2	Mohamed Al-Hussini	Hormone stimulation treatment for infertility in Klinefelter Syndrome - patient information
Р3	Mohamed Al-Hussini	Psychosexual issue affecting men with Klinefelter Syndrome seeking fertility management
P4	Sacha Salamon White	Risk of thromboembolism, and testosterone therapy in KS patients
Р5	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	Susan Howell	Eosinophilic esophagitis in individuals with sex chromosome aneuploidies: Clinical presentations and management implications
P8	Chaira Serrarens	Resting-state functional connectivity in adult women with 47,XXX: a 7 Tesla MRI study
Р9	Margo Willems	Identifying KS-specific fibrotic genes by analyzing transcriptome of (non-)fibrotic testicular tissue
P10	Alex Murray	Description of Klinefelter sample
P11	Matteo Spaziani	The dose-effect of supernumerary X chromosomes on clinical, metabolic and cardiac outcomes of male subjects
P12	Francesco Carlomagno	Altered thyroid feedback loop in Klinefelter syndrome: a longitudinal study from infancy, through transition-age to adulthood
P13	Simon Chang	Increased complement activation in men with Klinefelter syndrome
P14	Jörg Gromoll	Supervised machine learning based prediction models for identification of Klinefelter Syndrome
P15	Jesper Just	Adopting machine learning techniques to describe the phenotype of Klinefelter Syndrome using multiple categories of clinical measurements
P16	Megan Louderman	Temperament of very young children with SCT
P17	Margherita Vecchio	A review of the relationship between Klinefelter Syndrome and Gender Dysphoria
P18	Talia Thompson	Educational Supports for Children With Sex Chromosome Aneuploidies in the U.S.
P19	Kimberly Kuiper	Emotional reactivity and expressivity in young children with sex chromosome trisomies: evidence from psychophysiological and observational data
P20	Sophie van Rijn	The early impact of Sex Chromosome Trisomy (47XXX, 47XXY, 47XYY) on social cognition: evidence from eyetracking
P21	Rebecca Wilson	The eXtraordinarY Babies Study: Early Social Communication Skills in Infants with Sex Chromosome Trisomy (SCT)
P22	Evelien Urbanus	Social communication in children with sex chromosome trisomy (XXY, XXX, XYY): How children attend and respond to short communicative interactions
P23	Cristina Boada	The eXtraordinarY Babies Study: Early Adaptive Skills Profile of Infants and Toddlers with Prenatally Identified Sex Chromosome Trisomies
P24	Maarten Otter	Psychiatric symptoms in women with triple X-syndrome (TXS)
P25	Talia Thompson	An International Survey of Academic and Character Strengths in Youth with Sex Chromosome Aneuploidies
P26	Talia Thompson	Parenting young daughters with prenatally identified trisomy X
P27	Nienke Bouw	Early preventive intervention for young children with Sex Chromosome Trisomies (XXX, XXY, XYY)

