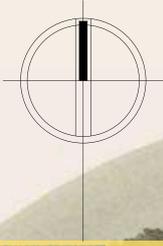


PADUA BOTANICAL GARDEN

UNIVERSITY OF PADOVA – UNESCO WORLD HERITAGE SITE

8 - 10 OCTOBER 2025



4th

INTERNATIONAL WORKSHOP ON

KLINFELTER SYNDROME

*and other supernumerary sex chromosome
aneuploidies*

**"A LIFELONG JOURNEY:
NEW ANSWERS TO OLD QUESTIONS"**

International Scientific Committee

Alberto Ferlin, Italy
Claus Gravholt, Denmark
Anders Juul, Denmark
Armin Raznahan, USA
Anne Skakkebak, Denmark

Hanna Swaab, The Netherlands
Nicole Tartaglia, USA
Sophie van Rijn, The Netherlands
Joachim Wistuba, Germany



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ENDORSEMENT



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INFORMATION

REGISTRATION FEES

The Registration includes **Scientific Materials, Coffee Breaks, Lunches** and **Welcome Dinner**.

€450,00 Standard Registration (*M.D.-all disciplines-, Psychologist, Biologist, Nurse*)

€250,00 Residents/Students/PhD Students

€100,00* Patients/Member of Patient Association

**This registration category does not include Scientific Materials and Welcome Dinner*

For Registration please click on the button below.

SIGN UP NOW



VENUE

Botanical Garden, University of Padova, via Orto Botanico 15, 35123 Padova. UNESCO World Heritage Site, the world's oldest University Botanical Garden in the world (1545).

HOW TO REACH PADUA BOTANICAL GARDEN

By Plane: Venice Marco Polo airport (VCE) is about 40 km far from Padua (Padova in Italian) – the city centre can be easily reached by taxi or public transport.

By Train: The railway station is close to the city centre. You can get to the Botanical Garden by bus or tram (leaving every 10 minutes from the railway station – stop n. 12-Santo).

By Car: There are parking facilities surrounding Prato della Valle.

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SCIENTIFIC RATIONALE

The 4th International Workshop on Klinefelter syndrome and other supernumerary sex chromosome aneuploidies with the theme "A lifelong journey: new answers to old questions" will be held on 8-10 October 2025 at the Padova Botanical Garden, University of Padova – UNESCO World Heritage Site. Previous international workshops were organized in Copenhagen (2010), Munster (2016), and Leiden (2022).

In this 4th International Workshop we will have different themes in a lifelong perspective, addressing the impact of an extra X or Y chromosome throughout life, with particular focus at critical periods, from birth, to infancy, adolescence, adulthood, and senescence. Attention will be given to the principal components of the syndromes and gonadal function, as well as to comorbidities, combining a comprehensive view of genetic, endocrine, metabolic, psychologic, social, and relational aspects, all participating in wellbeing and quality of life. Basic and translational research will be combined to more clinical and social issues, with also discussion on future challenges and new international guidelines.

Renowned experts, together with the International Scientific Committee, will present keynote presentations, and selected communications will be presented as Golden oral presentations and Oral communications, and in Poster session. Patients and patient associations will also be present.

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SCIENTIFIC PROGRAM

DAY 1 - WEDNESDAY OCTOBER 8TH

11:00-11:30 Opening and introduction

SESSION I: EPIDEMIOLOGY AND ETIOLOGY

Chairs: *Alberto Ferlin (Italy) – Hanna Swaab (the Netherlands)*

11:30-11:55 Epidemiology: do we really know the incidence and prevalence? – *Andrés Ingason (Denmark)*

11:55-12:20 Insights on Novel Genetic and Molecular Events at the Origin of Klinefelter Syndrome
– *Marco Barchi (Italy)*

12:20-12:30 **Golden Oral Presentation 1:** 15-year trends in US pediatric sex chromosome trisomy diagnoses –
Shanlee Davis (USA)

12:30-12:45 Discussion

12:45-14:00 Posters and lunch

SESSION II: GENOTYPE-PHENOTYPE RELATION

Chairs: *Nicole Tartaglia (USA) – Joachim Wistuba (Germany)*

14:00-14:25 Structure and features of the supernumerary X chromosome and the role of epigenetics – a stem cell
perspective - *Antonio Adamo (Saudi Arabia)*

14:25-14:50 Genetic counselling in XXY, XYY and Trisomy X and parent perspectives – *Susan Howell (USA)*

14:50-15:00 **Golden Oral presentation 2:** Congenital malformations in Klinefelter syndrome: A population-based
registry study – *Inger Dorf (Denmark)*

15:00-15:15 Discussion

15:15-16:00 Open Coffee break

Chair: *Anders Juul (Denmark)*

15:15-15:40 Milestones and developmental trajectories in sex chromosome trisomies – *Nicole Tartaglia (USA)*

15:40-15:50 Discussion

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SESSION III: WHAT REALLY HAPPENS TO TESTIS FUNCTION?

Chairs: *Armin Raznahan (USA) – Anne Skakkebaek (Denmark)*

15:50-16:15 Molecular and cellular events in the germ and somatic cells of the testis: animal models and human correlates – *Joachim Wistuba (Germany)*

16:15-16:40 Testicular microvasculature and inflammation – *Emma B. Johannsen (Denmark)*

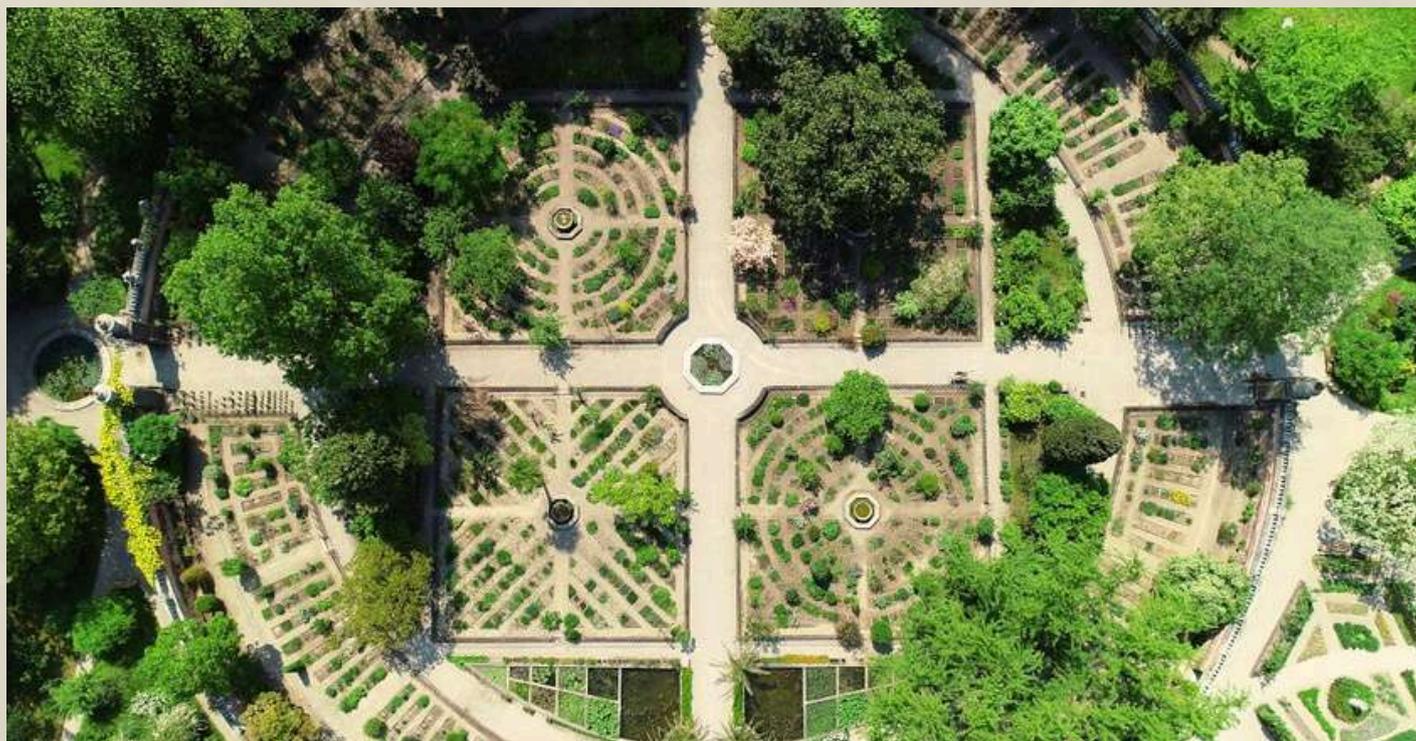
16:40-16:50 **Golden Oral presentation 3:** Timing of germ cell loss in non-mosaic Klinefelter Syndrome – *Semir Gül (Turkey)*

16:50-17:05 Discussion

Chair: *Linda Vignozzi (Italy)*

17:05-17:30 KING: the Klinefelter ItaliaN Group of the Italian Society of Andrology and Sexual Medicine (SIAMS) - *Francesco Carlomagno (Italy)*

20:00 Welcome dinner



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SCIENTIFIC PROGRAM

DAY 2 - THURSDAY OCTOBER 9TH

DEBATE I: MISSION: TO REDUCE UNDER-DIAGNOSIS

Chair: *Manuela Simoni (Italy)*

09:00-09:20 Is universal screening a solution? – *Claus Gravholt (Denmark)*

09:20-09:40 Knowledge, information and correct application of testing indications – *Giovanni Corona (Italy)*

09:40-10:00 Discussion

SESSION IV: HYPOGONADISM AND SPERMATOGENESIS

Chairs: *Carlo Foresta (Italy) – Andrea Isidori (Italy)*

10:00-10:25 When to start testosterone treatment – *Lise Aksglaede (Denmark)*

10:25-10:50 When and how to manage fertility issues – *Andrea Garolla (Italy)*

10:50-11:00 **Golden Oral presentation 4:** Stimulating the androgenic response of Leydig cells through administration of an LHR agonist to 41,XXY* male mice, a model for Klinefelter Syndrome –
Fariba Saadati (Germany)

11:00-11:15 Discussion

11:15-12:00 Open Coffee break

ROUND TABLE I: ISSUES IN CLINICAL CARE PRACTICE

Chairs: *TBC – Daniela Pasquali (Italy)*

11:15-11:35 Testosterone in infants – *Shanlee Davis (USA)*

11:35-11:55 Neurodevelopmental phenotypes of children with sex chromosome trisomies – *Hanna Swaab (the Netherlands)*

11:55-12:15 Transition from adolescence to adulthood – *Anders Juul (Denmark)*

12:15-12:30 Discussion

12:30-14:00 Posters and lunch

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DAY 2 - THURSDAY OCTOBER 9TH

ORAL COMMUNICATIONS

7' speech / 3' Q&A

Chairs:

Hanna Swaab (the Netherlands) – Armin Raznahan (USA)

14:00-14:10

Oral communication 1: Supernumerary X chromosomes shape brain organoid architecture and functions in a dose-dependent fashion – *Antonio Adamo (Saudi Arabia)*

14:10-14:20

Oral communication 2: Cardiometabolic evaluation in patients with Klinefelter Syndrome before and after testosterone replacement therapy – *Andrea Graziani (Italy)*

14:20-14:30

Oral communication 3: Serum estradiol (E2) and E2 to Total Testosterone (TT) ratio do not differ between patients with Klinefelter Syndrome (KS) and non-KS hypogonadism – *Daniele Renda Livraghi (Italy)*

14:30-14:40

Oral communication 4: DNA hypomethylation at specific CG-sites within TRAK1 is linked to the neurocognitive profile in Klinefelter syndrome – *Anne Skakkebaek (Denmark)*

14:40-14:50

Oral communication 5: Klinefelter syndrome and gender diversity in clinical practice – *Maaïke Kroon (the Netherlands)*

14:50-15:00

Oral communication 6: Testosterone replacement therapy drives multicellular remodeling of skeletal muscle in hypogonadal men with Klinefelter Syndrome – *Jesper Just (Denmark)*

15:00-15:10

Oral communication 7: Overall prevalence of Klinefelter Syndrome (KS), according to invasive prenatal diagnosis (IPD) procedures, its changes over time and geographical differences across the world: a systematic review – *Francesco Costantino (Italy)*

15:10-15:20

Oral communication 8: Social functioning as a predictor of development in adult women with triple x syndrome – *Maarten Otter (the Netherlands)*

15:20-15:30

Oral communication 9: Regulation of free fatty acid metabolism in Klinefelter syndrome – *Line Balsby Valsted (Denmark)*

15:30-15:40

Oral communication 10: Vascular Ageing in Conditions Associated With Early Onset Hypogonadism – *Angela Lucas-Herald (UK)*

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DAY 2 - THURSDAY OCTOBER 9TH

DEBATE II: DETERMINANTS OF WELLBEING AND QUALITY OF LIFE

Chair: *Nicole Tartaglia (USA)*

15:45-16:05 Sleep disorders in children and adolescents – *Luigi Tarani (Italy)*

16:05-16:25 Social, psychological and relational aspects – *TBC*

16:25-16:45 Discussion

16:45-17:30 Open Coffee break

SESSION V: NEUROLOGIC, COGNITIVE AND PSYCHOLOGIC ISSUE

Chairs: *Anne Skakkebaek (Denmark) – Hanna Swaab (the Netherlands)*

16:45-17:10 Trajectories of speech-language development in Klinefelter syndrome and other sex chromosome aneuploidies – *Laura Zampini (Italy)*

17:10-17:35 Neuropsychiatric outcomes in XXY, XYY and Trisomy X – *Armin Raznahan (USA)*

17:35-17:45 **Golden Oral presentation 5:** eXtraordinary Babies Study: A longitudinal study of early executive functioning skills in toddlers with sex chromosome aneuploidy – *Jennifer Janusz (USA)*

17:45-18:00 Discussion



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DAY 3 – FRIDAY OCTOBER 10TH

SESSION VI: COMORBIDITIES AND THEIR TREATMENT ACROSS THE LIFESPAN

Chairs: *TBC – Anders Juul (Denmark)*

09:00-09:25 The role of TRT on cardiometabolic risk and mortality – *Simon Chang (Denmark)*

09:25-09:50 Aging with Klinefelter syndrome and bone health – *Vincenzo Rochira (Italy)*

09:50-10:00 **Golden Oral presentation 6:** Psychiatric morbidity in Klinefelter syndrome: A population-based registry study – *Anna Colding (Denmark)*

10:00-10:15 Discussion

ROUND TABLE II: PHENOTYPE VARIATIONS BEYOND GENETICS AND TESTOSTERONE

Chairs: *Hanna Swaab (the Netherlands) – Joachim Wistuba (Germany)*

10:15-10:35 The transcriptome and epigenome across different tissues – *Anne Skakkebaek (Denmark)*

10:35-10:55 The role of familial environment in behavioral and neurocognitive phenotypes – *Sophie van Rijn (the Netherlands)*

10:55-11:15 The importance of the age at diagnosis and advantages of early diagnosis – *Alberto Ferlin (Italy)*

11:15-11:30 Discussion

11:30-12:00 **Open Coffee break**

CONCLUDING REMARKS

Chair: *Alberto Ferlin (Italy)*

11:30-11:50 Basic science research challenges – *TBC*

11:50-12:10 Clinical science research challenges – *Andrea Isidori (Italy)*

12:10-12:30 Guidelines – *Shanlee Davis (USA)*

12:30-12:45 General discussion

12:45-13:15 Conclusion and farewell



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ORGANIZING SECRETARIAT

For any further information
please contact:

THE ORGANIZING SECRETARIAT

CONGRESS  PLANNING

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