

5th I-DSD Symposium

Ghent, Belgium

2015



The New Zebra, Ghent
11th-13th June 2015

5th I-DSD Symposium Programme. 11th-13th June 2015, Ghent, Belgium

Welcome Remarks

Dear Friends and Colleagues,

On behalf of the I-DSD steering committee, it is with great pleasure that we welcome you to Ghent for the 5th International Symposium on Disorders of Sex Development from the 11th to 13th June 2015.

Once started as a German initiative, with the first two meetings held in Lübeck in 2004 and 2006, a third meeting was held in 2011 under the EuroDSD banner. Also in 2011, the UK Medical Research Council funded a partnership project (I-DSD) which was aimed at sustaining and extending the partnerships developed through the EuroDSD project. The cornerstone to this collaboration is the I-DSD Registry which has registered users from all corners of the world and is fast developing into a professional and scientific network. In addition, I-DSD maintains honoring the tradition of bringing together involved researchers and clinicians every two years at a top-level international scientific meeting, the first one held in Glasgow in 2013.

All these meetings have been very successful in attracting a rapidly expanding multidisciplinary group of experts, eager to discuss the latest advance in the field and have laid the foundation for fruitful international collaborations, such as the FP7 project Euro-DSD and the currently running COST Action BM1303, both funded by the EU. Along the way, patient representatives have increasingly become partners in setting the standards for the DSD-related research and care of the 21st century.

In a truly European spirit, this fifth meeting in Ghent is a joint organization of the DSD teams of Glasgow, Rotterdam and Ghent. A decade after the Chicago consensus meeting, the program has been developed to look to the future, encompassing basic and translational research, clinical care, psychological well-being and patient empowerment. The meeting will consist of invited speakers and original communications and there will be a workshop for investigators who are interested in performing clinical research in DSD and for those who would like to use the I-DSD Registry. There will be prizes for the best original communications and posters, sponsored by Karger. Thanks to a generous grant from the Ghent University, we have been able to award travel grants for professionals from low-income countries and patient representatives.

Ghent, elected by Lonely Planet as Europe's best hidden jewel in 2011, has a long standing tradition of freedom, humanism and skepticism against the established order. Beautifully enlightened at night, its medieval center, hosting numerous museums, churches and squares filled with restaurants and bars testifies of a glorious history and rich culture, with on top a taste of typical Belgian gastronomy and "joie de vivre". Trendy and cosmopolitan today, the city is one of the top Belgian destinations.

The meeting venue is a recently renovated building, aiming to promote modern art as well as social reintegration and offers apart from the most modern facilities a splendid terrace, on which we hope you can make plans for your future collaborations under an inspiring, although somewhat capricious Belgian sunshine. As impressive but from another world, we hope to welcome you on Friday night for our social dinner in the ancient abbey of Sint Pieter.

We would like to acknowledge the generous support of the Medical Research Council UK. As organizing committee we look forward, together with you, to an enriching meeting, meant to be a stimulating scientific and friendly environment, where initiatives can be initiated to further increase quality of understanding and translation to care.

Faisal Ahmed, Jillian Bryce, Leendert Looijenga and Martine Cools

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General Information

Meeting Venue:	The New Zebra Entrance on Gustaaf Callierlaan 232 (also Zebrastraat 32)
Workshop Venue:	The New Zebra Entrance on Gustaaf Callierlaan 232 (also Zebrastraat 32)
Internet:	Zebrapoint (no password required)
Badges:	Name badge upon registration. Permits access to Symposium Sessions and catering facilities. (Does not permit access to DSD workshop - separate registration required)
Meals:	Lunches and coffee breaks included in registration fee
Social Events	Entry by pre-ordered ticket only - supplied in registration pack.
Conference Dinner:	Sint Pieters Abdij , Sint Pietersplein 9, 9000 Ghent, Friday 12 th June, 8pm
Excursion:	The Medieval City of Ghent , Green Boathouse, Korenlei 4A, 9000 Ghent, Saturday 13 th June, 2pm
Accreditation:	UEMS-EACCME for 15 hours CME. Compatible with USA and Canadian CME schemes. CME Certificates of Attendance will be provided at the Symposium upon completion of a feedback survey.
Organising Committee:	Martine Cools, Leendert Looijenga, An Desloovere (Ghent/Rotterdam) Faisal Ahmed, Jillian Bryce (Glasgow)
Additional Support:	Special thanks to the volunteers from Ghent University for the additional ground support
Supported by:	MRC UK Ghent University University of Glasgow COST-BM1303 DSDnet Karger (Sexual Development)
Supported by:	



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Symposium Day 1 (Thursday 11/6/2015)

I-DSD Training Workshop			
8:30-9:00 Entrance Hall	Registration		
9:00-10:45 Nick Ervinck Panamarenko Thomas Huyghe	Use of the registry J Jiang, M Rodie (University of Glasgow)	E-learning S Drop (Erasmus MC)	Communication skills C Quigley (Indiana University) E Magritte (dsdfamilies.org)
10:45-11:00 Lounge	Coffee break (15')		
11:00-12:00 Nick Ervinck Panamarenko Thomas Huyghe	Use of the registry	E-learning	Communication skills
12:00-12:45 Lounge	Workshop Lunch		

I-DSD Symposium	
11:00-17:00 Entrance Hall	Registration
12:50-12:55 Ned Khan	M Cools (Ghent University): Opening and welcome
12:55-15:00 Ned Khan	10 Years After Chicago - a Look to the Future Chair: Olaf Hiort (University of Lübeck)
12.55–13.00	M Muscarella (dsdteens.org, dsdfamilies.org, Cambridge MA): Setting the scene for the session
13.00–13.20	I Hughes (University of Cambridge): The Chicago Consensus and its consequences
13.20–13.40	C Sanders (Alder Hey Hospital, Liverpool): Supporting the affected child and family
13.40–14.00	J Achermann (University College London): A decade of diagnosing DSD conditions
14.00–14.20	P Hoebeka (University of Ghent): Genital construction and its timing
14.20–14.40	F Ahmed (University of Glasgow): The I-DSD Registry and related networks
14.40–15:00	Discussion
15:00-15:30 Ned Khan bar	Coffee Break (30')

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15:30-16:05 Ned Khan	Short Oral Communications 1 Chair: Anna Nordenström (Karolinska Institute, Stockholm)
5'	A Kyriakou (I-DSD/DSDnet): Current models of practice & professional development of clinicians in DSD centres - results from an international survey of specialist care for DSD N Callens (University of Michigan): A web-based tool for shared decision-making in DSD. A Morris (University of Melbourne): Congenital adrenal hyperplasia(CAH) in Melbourne: Surgical timing and complications, with outcomes including body image and genital sensation in a cohort study M Rodie (Scottish DSD Network): The Scottish audit of atypical genitalia TC van de Grift (Vrije Universiteit, Amsterdam): Well-being of people with an intersex condition - a qualitative study on experiences with medical care and quality of life. R Loch Batista (University of São Paulo): Gender identity of prediction in old age by htp test (house-tree-family) in subjects with DDS 46, XY T Lundberg (University of Oslo): From knowing nothing to "Knowing-now": Experiences and expertise among parents of children with CAH
16:05-16:15	Short Pause (10')
16:15-17:00 Ned Khan	Short Oral Communications 2 Chair: Inas Mazen (National Research Centre, Cairo)
5'	V Harley (MIMR-PHI Institute of Medical Research): Unraveling embryonic testicular pathways via SOX9 KO RNAseq and SOX9 ChIPseq MM Rutter (University of Cincinnati): A tale of two sisters G Russo (University "Vita e Salute"): 5alpha-reductase-2 deficiency: clinical findings, endocrine profile and genetic features in 25 persons E Vilain (University of California Los Angeles): Reducing uncertainty: Exome sequencing for the diagnosis of 46,XY disorders of sex development. H Özbeyp (Istanbul Medical Faculty): Morphological characteristics of the gubernaculum in undescended testes N Dessouky (Children's University Specialized Hospital, Cairo): Ovotesticular disorders of sex development Surgical diagnosis and management of 32 cases A Titmuss (Children's Hospital at Westmead): Gonadal pathology in 45,X/ 46,XY females M Cools (Ghent University): Prediction of germ cell cancer occurrence in postpubertal individuals with androgen insensitivity EH Quint (University of Michigan): Gonadectomy outcomes in patients with 46,XY DSD
17:00-17:50 Ned Khan	Opening Guest Lecture Chair: Faisal Ahmed (University of Glasgow)
17:00-17:05	Introduction of Guest Speaker
17:05-17:40	Claudia Wiesemann (University of Göttingen): The ethics of medical care for children with DSD
17:40-17:50	Discussion
	Free Evening

Symposium Day 2 (Friday 12/06/2015)

<i>Parallel Sessions</i>			
9:00-10:30 Ned Khan	The Biology of Sex Development Chair: Ken McElreavey (Institut Pasteur)	Processes, Not Events: Achieving Better Longterm Outcomes Host: Family and Patient Groups	9:00-10:30 Nick Ervinck
9.00-9.30	P Koopman (University of Queensland): SRY: from basic genetics to complex epigenetics	E Magritte (dsdfamilies) & J Gorter-Bouma (dsdnederlanden): From I-DSD Glasgow to I-DSD Ghent: from 'patient groups' to 'partners'?	9:00-9:05
9.30-10:00	D Zarkower (University of Minnesota): Plasticity of the gonad	T Devore (Hypospadias/Epispadias Association): Working together in raising confident young men	9:05-9:30
10:00-10.30	G Yamada (Wakayama Medical University): Development of the External Genitalia	L-M Liao (UCLH): Informed consent: from ethical principles to ethical behaviour Various invited speakers: Building partnerships to enhance health literacy and facilitate peer-to-peer support.	9:30-9:55 9:55-10:30
10:30-11:00 Ned Khan bar	Coffee Break (30')		
11.00-12.30 Ned Khan	Long Oral Communications Chair: Amy Wisniewski (University of Oklahoma) & Laura Audi (Hospital Vall d'Hebron)	Clinical Psychology: Stress and Distress in DSD Chair: A Dessens (ERASMUS MC) & E Van Hoecke (Ghent University)	11:00-12:30 Nick Ervinck
11:00-11:10	A Sinclair (Murdoch Children's Research Institute): Massively parallel sequencing of a targeted panel for the diagnosis of DSD	E van Hoecke (Ghent University) Welcome and introduction	11:00-11:05
11:10-11:20	S Flieger (University of Lübeck): A new Next Generation Sequencing panel for mutational screening of putative genes causing 46, XY DSD	V Pasterski (University of Cambridge): Posttraumatic stress in parents of children diagnosed with DSD - research findings and its clinical implications	11:05-11:25
11:20-11:30	D Baetens (Ghent University): Identification and functional characterization of ESR2, a new disease gene for 46,XY DSD	C Sanders (Alder Hey Hospital): Parental narratives about their child's genital ambiguity – protection and decisions on genital surgery	11:25-11:40
11:30-11:40	AZ Juniarto (Diponegoro University): Hormonal evaluation in relation to phenotype and genotype in 286 patients with a DSD from Indonesia	A Dessens (Erasmus MC) & A Ediati (Semarang): Emotional distress in Indonesian patients with DSD	11:40-11:55
11:40-11:50	A Kyriakou (DSDnet): Diagnostic approach to a newborn with suspected DSD - results from an international survey of specialist care for DSD	B Stockman & B Van Hoorde (University of Ghent): Multidisciplinary approach in vaginal dilation therapy General Discussion How to reduce the stress? What type of stress management is needed? How to comfort the distressed?	11:55-12:10 12:10-12:30

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11:50-12:00	A Lucas-Herald (University of Glasgow): Long-term endocrine outcome in men with partial androgen insensitivity syndrome	
12:00-12:10	MC Meriggiola (University of Bologna): Bone mineral density in women with complete androgen insensitivity syndrome: effects of gonadal removal and sex steroids substitutive therapy	
12:10-12:20	J van Hemmen (Netherlands Institute for Neuroscience): Brain white matter microstructure in women with CAIS	
12:20-12:30	M Szarras-Czapnik (Children's Memorial Health Institute, Warsaw): Neoplastic risk of dysgenetic testes	
12:30-13:30 Lounge	Lunch with idea drop box	
13:30-15:20 Ned Khan	The Complexities of Phenotypic Assessment Chair: Rodolfo Rey (Hospital de Niños Ricardo Gutiérrez)	
13.30–14:00	A Springer (University of Vienna): Comprehensive clinical phenotyping of DSD	
14:00–14:30	C Flück (University of Bern): Sex steroid synthesis	
14.30–15.00	S Mueller (University of Ghent): Neural correlates of steroid disorders	
15:00-15.20	General discussion	
15:20-16:00 Ned Khan bar	Coffee Break with idea drop box (40')	
16.00–17.00 Gallery	Poster session	
17.00–17.45 Ned Khan	Discussion forum based on pre-identified topics and content of idea drop box. Chair: Leendert Looijenga (Erasmus Medical Center, Rotterdam)	
20:00 Sint Pieters Abdij	Social evening (Conference dinner)	

Symposium Day 3 (Saturday 13/06/2015)

9:00-10:30 Ned Khan	The Gonad and its Long-Term Outcome Chair: Guy T'Sjoen (Ghent University Hospital)
09.00-09:30	O Söder (Karolinska Institute, Stockholm): Endocrine disruptors and their effects on gonadal function
09.30-10.00	L Looijenga (Erasmus Medical Centre, Rotterdam): GCC risk profiles: Relevance for DSD
10.00-10:30	M Johansen (University of Copenhagen): Long-term outcome with a focus on growth, sexual development and fertility
10:30-11:00 Ned Khan bar	Coffee break (30')
11:00-12:00 Ned Khan	Care Across the Age Span Chair: Berenice Mendonca (University of Sao Paolo)
11.00-11.20	N Callens (University of Michigan, Ann Arbor): Vaginal hypoplasia in adolescents
11.20-11.40	K Schenk-Gustafsson (Karolinska Institute, Stockholm): Sex hormones & cardiovascular health
11.40-12.00	J Pippi Salle (Sidra Medical Centre, Doha) Long term problems after genitoplasty in males, importance of follow-up
12:00-13:00 Ned Khan	Closing Guest Lecture Chair: Piet Hoebeke (Ghent University Hospital)
12.00-12:05	Introduction of Guest Speaker
12:05-12.35	Marleen Temmerman (WHO, Geneva): Sexual health, human rights and law
12:35-12:45	Discussion
12:45-12:50	Closure of the meeting (Leendert Looijenga, Faisal Ahmed)
14:00-15:00 Green boathouse	Excursion (Boat Trip in Ghent)

Symposium Day 2 (Friday 12/06/2015)

Poster Presentations

Gallery, New Zebra

Posters will be displayed on Friday 12th June in the Gallery. Posters should be mounted in the morning and removed after the end of the attended poster session (4-5pm).

P1 Communication With Peer Support Groups And Families - Results From An International Survey Of Specialist Care For DSD. Marta Rozas, Support Group GrApSIA, Barcelona, Spain

P2 Follow-Up Studies: The Good, The Bad, and The Ugly. Michael Kreuzer, University of Ottawa, Canada

P3 Sex Testing of Elite Female Athletes with DSD: Science and Controversies. Maria José Martinez-Patiño, University of Vigo-Institute of Biomedical Research (IBI-Uvigo), Spain

P4 Incorporating Support & Advocacy into the Disorders of Sex Development – Translational Research Network (DSD---TRN): Building Relationships and Resources for Patients, Families, and Healthcare Providers. Janet Green, Accord Alliance/DSD-TRN

P5 Applying Hume's Is/Ought Problem to 'Disorder of Sex Development. Natalie Delimat, Institute of Technology, Sligo, Ireland.

P6 Body Image and Quality of Life (QoL) in women with Congenital Adrenal Hyperplasia - outcomes and avenues for adjuvant treatment . Erika Tomlinson, Austin/Northern Health, Melbourne, Australia

P7 Psychosocial well-being in Dutch adults with a disorder of sex development. N.G.M. de Neve-Enthoven, Erasmus Medical Center Rotterdam, Netherlands

P8 Psychological consequences for DSD patients with Y chromosome. Katarzyna Bajszczak, Children's Memorial Health Institute, Warsaw, Poland

P9 Health care-seeking behavior in late-identified patients with congenital adrenal hyperplasia (CAH) in Central Java, Indonesia. Agustini Utari, Diponegoro University, Indonesia

P10 The body image and genitalia appearance concerns and virilization features of CAH female patients in Malaysia. Ani Amelia Zainuddin, The National University of Malaysia (UKM), Kuala Lumpur, Malaysia

P11 Islamic perspectives of DSD and gender-related issues. Ani Amelia Zainuddin, The National University of Malaysia (UKM)

P12 "We are also humans we who are sick" Experiences of 13 women with congenital adrenal hyperplasia. Hedvig Engberg, Karolinska Institutet and Child and Adolescent Psychiatry Research Center, Stockholm, Sweden

P13 Multidisciplinary and psychosocial shift in DSD/Intersex management. Nuria Gregori Flor, Valencia University, Spain

P14 Three novel CYP21A2 mutations identified in Brazilian patients with 21-Hydroxylase deficiency: A synergistic effect. Débora de Paula Michelatto, Karolinska University Hospital, Sweden

P15 FGFR2 mutation in XY sex reversal with craniosynostosis. Vincent Harley, MIMR-PHI Institute of Medical Research, Melbourne, Australia

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P16 FGF9 activity from normal males and a 46,XY female. Vincent Harley, MIMR-PHI Institute of Medical Research, Melbourne, Australia

P17 Characterization of mutations in the androgen receptor (AR) identified in 38 Brazilian families with complete or partial androgen insensitivity syndrome (AIS). Rafael Loch Batista, University of São Paulo, São Paulo-SP, Brazil

P18 Sex chromosomal Abnormalities in Egyptian DSD patients. Inas Mazen, National Research Center, Cairo, Egypt

P19 Functional Analysis of three NR5A1 mutations identified in patients with 46,XY DSD. Helena Campos Fabbri, State University of Campinas, Brazil

P20 New case of androgen insensitivity syndrome due to A645D mutation of AR gene associated with short polyglycine and long polyglutamine repeats. Rita Ortolano, University of Palermo, Italy

P21 Chromosomal and molecular studies in a cohort of 88 patients with Disorders of Sex development. Silvia Majore, San Camillo-Forlanini Hospital, Rome, Italy

P22 Gender Re-assignment of a 46,XY-DSD Patient Associated with P450 Oxidoreductase Gene (POR) Mutation and Antley-Bixler Syndrome. Hüseyin Özbeý, Istanbul University & DSDTurk, Istanbul, Turkey

P23 A novel WT1 gene mutation that affects the splicing only of the - KTS isoform identified in a boy with 46,XY DSD and normal renal function. Lilia Baldazzi, University of Bologna, Bologna, Italy

P24 The Fuzziness of Next Generation Sequencing Analysis: A Case Study on DSD Patients in Australia. Richard Sinnott, University of Melbourne, Australia

P25 Diagnostic approaches of disorders of sex development (46, XY DSD) by genetic methods. Azadeh Shojaei, Iran University of Medical Sciences, Tehran, Iran.

P26 Molecular analysis of the AR gene in two patients with 46,XY CAIS - a recurrent (p.Agr616His) and a novel (p.Ile842Asn) mutations. Silvia Andonova , Medical University Sofia, Bulgaria

P27 Syndromic DSD; A study of 30 Egyptian cases using cytogenetic techniques.. Mona A. El Gammal, Clinical Genetics Department, National Research Centre, Cairo, Egypt

P28 Phenotypic variability associated with NR5A1 gene mutations. Idoia Martinez de LaPiscina, CIBERDERM, Barakaldo, Spain

P29 Expression of Gonadoblastoma Y (GBY) locus in germ cells of gonads from DSD-XY female patients: risk assessment for occurrence of gonadoblastoma & dysgerminoma. Peter H. Vogt, University of Heidelberg, Germany

P30 Ovarian Adrenal Rest Tumour in a CAH patient. Ani Amelia Zainuddin, The National University of Malaysia (UKM)

P31 Long-term urinary symptoms in adolescent and adult women with congenital adrenal hyperplasia. Sonia R Grover, University of Melbourne, Australia

P32 Managing genital difference in children with anesthesia risks: an alternative approach. Arlene B. Baratz, Disorders of Sex Development Support Group, USA

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P33 Pubertal Development in Individuals with Partial Androgen Insensitivity Syndrome (PAIS) Assigned Female Sex of Rearing. Guilherme Guaragna-Filho, State University of Campinas, Brazil

P34 Management of gonads in adults with androgen insensitivity: an international survey. Ellen Mari, Ghent University Hospital, Belgium

P35 Prenatal diagnosis of DSD: Initial experience of a multidisciplinary team. S. Vallasciani, Ospedale Maggiore Policlinico, Milan, Italy

P36 Challenges in the management of affected children with different gender in a multiplex CAH family Delayed referral: challenges in the clinical management of CAH in Indonesia. Agustini Utari, Diponegoro University, Indonesia

P37 An anatomical hypospadias repair incorporating Y-V preputioplasty in distal urethral reconstruction. Hüseyin Özbeý, Istanbul University & DSDTurk, Istanbul, Turkey.

P38 Clinical features in three patients with 46,XY DSD NR5A1 related. Marrocco Giacinto, S. Camillo-Forlanini Hospital, Rome, Italy

P39 Multi-disciplinary and Novel Approach to Treatment of 46,XY Disorder of Sex Development. Courtney Finlayson, Northwestern University Feinberg School of Medicine, Chicago, USA

P40 A tale of three Sudanese sisters with congenital adrenal hyperplasia. Mona Ellaithi, Al-Neelain University, Khartoum and Ahfad University for Women, Omdurman , Sudan

P41 Development of a Decision-Making Checklist to Improve Care for Adolescents with Complete Androgen Insensitivity Syndrome (CAIS). Kathleen Graziano, Phoenix Children's Hospital, USA

P42 46XY ovotesticular DSD: what's the correct approach? Maria Grazia Scarpa, Institute for Maternal and Child Health,Trieste, Italy

P43 Herlyn-Werner-Wunderlich Syndrome with unusual presentation in a 15 years old girl. Vilhelm Mladenov, University Hospital "Sveta Marina", Varna, Bulgaria

P44 Rare case of familial XY complete gonadal dysgenesis. Radoslava Vazharova, Sofia University. "St Kliment Ohridski", Bulgaria

P45 Complete androgen insensitivity syndrome - a case report and new trends in the management. Zofia Kolesinska, Poznan University of Medical Sciences, Poland

P46 Newborns with Congenital Adrenal Hyperplasia and Markedly Virilized Genitalia: Considerations of Alternative Gender Assignments. Francisco Javier Blas,UCLA DSD

P47 A rare case of ovotesticular DSD associated with 46,XX/46,XY tetragametic chimerism. Yolande van Bever, Erasmus Medical Centre, Rotterdam, the Netherlands

P48 The Disorders of Sex Development Translational Research Network (DSD-TRN). Emmanuèle Délot, UCLA, Los Angeles

P49 Securing the I-DSD Registry. John Watt and the I-DSD Project Management Team, University of Glasgow, UK