

7th I-DSD Symposium Programme. July 2019, São Paulo, Brazil
Faculdade de Medicina da Universidade de São Paulo

I-DSD Training Workshop Thursday July 4th 2019 (09:00 -12:30) Classrooms 2266, 2368 & 2370, 2 nd floor Faculty of Medicine Rotation through 3 workshops lasting 45 mins each		
Angela Lucas-Herald & Salma Ali (Glasgow, UK) Using the I-DSD/CAH registries	Amit Pandey (Bern, Switzerland) Bioinformatic tools	Amy Wisniewski (Fort Worth, USA) Communication with a new parent
Lunch for workshop – Faculty of Medicine Cafeteria (Basement) 12:00 – 12:30		

Symposium Day 1 (Thursday, July 4th 2019)
All sessions in Faculty of Medicine Theatre

Registration & Coffee from 11:30 – Foyer	
13:00: Opening	
Session 1 – Setting the Scene (13:00-14:30)	
13:00 – 13:10	Berenice B. Mendonca (São Paulo, Brazil) & Faisal Ahmed (Glasgow, UK)
13:10 – 13:20	Patients’ experiences introduced by Guilherme Guaragna-Filho (Porto Alegre, Brazil)
Session 2 – The Impact of Collaborations Chairs: Anna Nordenström, Ivo Arnhold	
13:20 – 13:35	Impact of European collaboration - Olaf Hiort (Lübeck, Germany)
13:35 – 13:50	Impact of collaboration in North America - David Sandberg (Ann Arbor, USA)
13:50 – 14:05	A network for DSD in the Asia & Pacific Region - Reiko Horikawa (Tokyo, Japan)
14:05 – 14:20	I-DSD/I-CAH & EuRECa – a template for the future - Faisal Ahmed (Glasgow, UK)
14:20 – 14:30	Future Directions – Round-table
Coffee Break - 14:30 – 15:00 – Faculty of Medicine Cafeteria (Basement)	
15:00 – 16:30	Oral communications 1 (OC1) Chairs: Martine Cools, Richard Auchus
15:00 – 15:12	Pathogenic variants in the fourth zinc finger domain of Wilms’ tumor 1 (WT1) gene are associated with 46,XX testicular/ovotesticular DSD – Anu Bashambo (Paris, France)
15:12 – 15:24	The Mobile DNA at Work: L1 Retrotransposition in the 5’UTR of the Androgen Receptor Causing Partial Androgen Insensitivity Syndrome – Rafael Loch Batista (Sao Paulo, Brazil)
15:24 – 15:36	Development Of An International Benchmark For Sick Day Episodes As A Core Clinical Outcome In People With Congenital Adrenal Hyperplasia – Salma Ali (Glasgow , UK)
15:36 – 15:48	Feminizing Genitoplasty in Patients with Disorder of Sex Development. Evaluation of Long–term Results of Anatomy, Genital Sensibility, Sexual Function and Satisfaction with Surgical Results – Min Jeong Bag (Sao Paulo, Brazil)
15:48 – 15:55	Growth, pubertal course and long-term outcome of 46,XY boys born with atypical genitalia and low birthweight - Martine Cools (Ghent, Belgium)

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15:55 – 16:02	A Nationwide Study Of The Prevalence & Initial Management Of Atypical Genitalia & Delayed Sex Assignment In The Newborn - Salma Ali (Glasgow, UK)
16:02 – 16:09	Endocrine and reproductive outcome of men born with various degrees of hypospadias - Martine Cools (Ghent, Belgium)
16:09 – 16:16	The Practice Of Gonadectomy In Cases Registered In The I-DSD Registry - Angela Lucas-Herald (Glasgow, UK)
16:16 – 16:23	Broad phenotypes of disorders/differences of sex development in MAMLD1 patients through oligogenic disease - Christa Flück (Bern, Switzerland)
16:23 – 16:30	Mutations in the DEAH-box RNA Helicase DHX37 are a frequent cause of 46,XY gonadal dysgenesis and 46,XY testicular regression syndrome – Ken McElreavey (Paris, France)
Short Break 16:30 – 16:40	
Session 3 – Lecture (16:40-17:20)	
Chair: Berenice B. Mendonca	
16:40 – 17:10	A short tribute to Claude Migeon Family-centered, interdisciplinary care: Educating DSD people and their families and loved ones - Amy Wisniewski (Fort Worth, USA)
17:10 – 17:20	Discussion

Free Evening

Symposium Day 2 (Friday, July 5th 2019)

Session 4 - Improving Diagnostic Outcomes in DSD (08:15-10:00)	
Chairs: Ana Latronico, Vincent Harley	
08:15 – 08:45	The strengths and pitfalls of genetics – Ken McElreavey (Paris, France)
08:45 – 09:15	The strengths and pitfalls of biochemistry – Richard Auchus (Ann Arbor, USA)
09:15 – 09:45	Combining genetics & biochemistry – Christa Flück (Bern, Switzerland)
09:45 – 10:00	Discussion
Coffee Break 10:00 – 10:30 - Faculty of Medicine Cafeteria (Basement)	
10.30 – 12:00	Oral communications 2 (OC2) Chairs: Alicia Belgorosky, Olaf Hiort
10:30 – 10:42	PPP2R3C Gene Variants Cause Syndromic 46,XY Gonadal Dysgenesis and Impaired Spermatogenesis in Humans – Tulay Guran (Istanbul, Turkey)
10:42 – 10:54	FGF9 missense variants affect testis development – Vincent Harley (Clayton, Australia)
10:54 – 11:06	Vascular dysfunction in children with hypospadias – Angela Lucas-Herald (Glasgow, UK)
11:06 – 11:18	Long-term urological and psychosexual outcome of men born with hypospadias – Martine Cools (Ghent, Belgium)
11:18 – 11:25	WES analysis of a cohort of 94 DSD patients - Anna Biason-Lauber (Fribourg, Switzerland)

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11:25 – 11:32	Gonadoblastoma Y (GBY) genes, DDX3Y and TSPY, are differentially expressed in the germ cells of DSD-46,XY individuals with CAIS and Swyer syndrome; these studies reveal UTY as third GBY candidate gene – Peter Vogt (Heidelberg, Germany)
11:32 – 11:39	Clinical, Hormonal, Psychosexual Aspects, and Genetic Characteristics of Androgen Insensitivity Syndrome in a Androgen Insensitivity Cohort: Long Term Outcomes and Novel Androgen Receptor Gene Allelic Variants - Rafael Loch Batista (São Paulo, Brazil)
11:39 – 11:46	Urethral Stricture In The Long-Term Follow-Up Of Patients With DSD Submitted To Hypospadias Repair - Min Jeong Bag (São Paulo, Brazil)
11:46 – 11:53	Reproductive Outcomes in Females with Virilizing Forms of Congenital Adrenal Hyperplasia – Tania Bachega (São Paulo, Brazil)
11:53 – 12:00	A Health-Related Quality of Life Tool for Parents of Young Children With Disorders of Sex Development - Salma Ali (Glasgow , UK)
Session 5 - Improving Health Outcomes in Sex Chromosome Disorders (12:00 - 13:30) Chairs: Amy Wisniewski, Durval Damiani	
12:00 – 12:30	Turner Syndrome – Theo Sas (Rotterdam, Netherlands)
12:30 – 13:00	Klinefelter Syndrome - Claus Gravholt (Aarhus, Denmark)
13:00 – 13:30	45X/46XY - Martine Cools (Ghent, Belgium)
Lunch 13:30 – 14:30 – Faculty of Medicine Cafeteria (Basement)	
13.40 - 14.20	Challenging case discussions in DSD Cases presented by Hedi Claahsen (Nijmegen, Netherlands) & Berenice Mendonca (São Paulo, Brazil) Moderators: David Sandberg & Rodolfo Rey
Session 6 - Congenital Adrenal Hyperplasia (14:30 - 16:20) Chairs: Christa Fleuck, Gil Guerra-Junior	
14.30 – 15.00	Diagnosis of CAH – Roxana Marino (Buenos Aires, Argentina)
15.00 – 15.30	Controversies in steroid replacement - Tania Bachega, (São Paulo, Brazil)
15.30 – 16.00	Critical outcomes in virilising CAH – Hedi Claahsen, (Nijmegen, Netherlands)
16:00 – 16:20	Discussion
Coffee and Posters 16.30 – Faculty of Medicine (Ground floor)	
17.00 – 18:30	Guided poster rounds
20.00 - Social Evening (Dinner 5th floor Faculty of Medicine with music by duo Bico de Pena)	

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Symposium Day 3 (Saturday July 6th 2019)

Session 7 - Improving Surgical Outcomes (09:00 – 10:30)	
Chairs: Eduardo Correa Costa, Steven Chernausek	
09:00 – 09:20	Female genitoplasty – Maria Helena Sircili (São Paulo, Brazil)
09:20 – 09:40	Male genitoplasty – Francisco Tibor Dénes (São Paulo, Brazil)
09:40 – 10:00	Postponing surgery in CAH: Nothing is ever black and white – Ignacio Bergadá (Buenos Aires, Argentina)
10:00 – 10:20	Auditing outcomes of surgery – Stuart O’Toole (Glasgow, UK)
10:20 – 10:30	Discussion
Coffee Break 10:30 – 11:00 – Faculty of Medicine (5th floor)	
Session 8 - This house believes that it is time to discard the term ‘DSD’ (11:00 – 12:00)	
Chairs: Berenice Mendonca & Faisal Ahmed	
11:00 – 11:15	For – Eric Vilain, (Los Angeles, USA)
11:15 – 11:30	Against – Andy Greenfield, (Oxford, UK)
11:30 – 12:00	Discussion
12:00 – Close (Faisal Ahmed)	



The 7th International DSD Symposium, São Paulo, Brazil, 04/07/2019-06/07/2019 has been accredited by the European Accreditation Council for Continuing Medical Education (EACCME®) with 13 European CME credits (ECMEC®s). Each medical specialist should claim only those hours of credit that he/she actually spent in the educational activity.

Feedback survey www.surveymonkey.co.uk/r/I-DSD2019

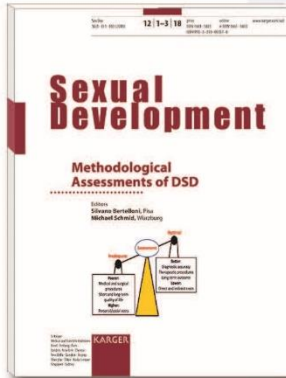
Abstracts home.i-dsd.org/i-dsd-symposium-2019/

#IDSDSYMPOSIUM



Essential reading on health care related to DSD

Methodological Assessments of DSD



Editors
S. Bertelloni
M. Schmid

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- The Role of Copy Number Variants in Disorders of Sex Development: **Croft, B.; Ohnesorg, T.; Sinclair, A.H.**
- Importance of Serum Testicular Protein Hormone Measurement in the Assessment of Disorders of Sex Development: **Freire, A.V.; Grinspon, R.P.; Rey, R.A.**
- Human Chorionic Gonadotropin Test: Old Uncertainties, New Perspectives, and Value in 46,XY Disorders of Sex Development: **Bertelloni, S.; Russo, G.; Baroncelli, G.I.**
- Normative Basal Values of Hormones and Proteins of Gonadal and Adrenal Functions from Birth to Adulthood: **Fanelli, F.; Baronio, F.; Ortolano, R.; Mezzullo, M.; Cassio, A.; Pagotto, U.; Balsamo, A.**
- Imaging Techniques in the Diagnostic Journey of Disorders of Sex Development: **Guerra-Junior, G.; Andrade, K.C.; Barcelos, I.H.K.; Maciel-Guerra, A.T.**
- Endoscopy and Laparoscopy in Disorders of Sex Development: **Tafazzoli, K.; Wünsch, L.; Bouteleux, M.; Lindert, J.; Schulz, T.; Birnbaum, W.; Marshall, L.; Hiort, O.; Tüshaus, L.**

- Histological Assessment of Gonads in DSD: Relevance for Clinical Management: **Spoor, J.A.; Oosterhuis, J.W.; Hersmus, R.; Biermann, K.; Wolffenbuttel, K.P.; Cools, M.; Kazmi, Z.; Ahmed, S.F.; Looijenga, L.H.J.**
- Methodological Issues for Psychological Evaluation across the Lifespan of Individuals with a Difference/Disorder of Sex Development: **D'Alberton, F.; Vissani, S.; Ferracuti, C.; Pasterski, V.**
- Improving Laboratory Assessment in Disorders of Sex Development through a Multidisciplinary Network: **Hannema, S.E.; de Rijke, Y.B.**
- Virtual Networks for Exchanging Information and Biomaterials: Future Directions: **Kourime, M.; Ahmed, S.F.**
- Multiparameter Investigation of a 46,XX/46,XY Tetragametic Chimeric Phenotypical Male Patient with Bilateral Scrotal Ovotestes and Ovulatory Activity: **van Bever, Y.; Wolffenbuttel, K.P.; Brüggewirth, H.T.; Blom, E.; de Klein, A.; Eussen, B.H.J.; van der Windt, F.; Hannema, S.E.; Dessens, A.B.; Dorssers, L.C.J.; Biermann, K.; Hersmus, R.; de Rijke, Y.B.; Looijenga, L.H.J.**
- Concluding Remarks - From Bench to Bed: **Hiort, O.**

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