Poster Presentations
1. Preserved bone mineral density in adults with classical congenital adrenal hyperplasia submitted to low-dose. Tania Bachega, Unidade de Adrenal, Laboratório de Hormônios e Genética Molecular LIM 42, Disciplina de Endocrinologia, Hospital

2. Efficacy of liquid-chromatography and radioimmunoassay in false-positives’ drop-off in CAH newborn screening. Tania Bachega, Unidade de Adrenal, Laboratório de Hormônios e Genética Molecular LIM 42, Disciplina de Endocrinologia, Hospital

3. Impact of long-term dexamethasone therapy on the metabolic profile of adult patients with classic forms of 21-hydroxylase deficiency. Tania Bachega, Unidade de Adrenal, Laboratório de Hormônios e Genética Molecular LIM 42, Disciplina de Endocrinologia, Hospital

4. Adverse outcomes and economic burden of congenital adrenal hyperplasia (CAH) late diagnosis in the absence of newborn screening. Tania Bachega, Unidade de Adrenal, Laboratório de Hormônios e Genética Molecular LIM 42, Disciplina de Endocrinologia, Hospital

5. Feminizing Genitoplasty in congenital Adrenal Hyperplasia: Results on the experience of a tertiary centre. Eduardo Correa Costa, Hospital de Clinicas de Porto Alegre

6. Gender Dysphoria and XX congenital adrenal hyperplasia: how frequent is it? Is Male-Sex rearing a good idea? Eduardo Correa Costa, Hospital de Clinicas de Porto Alegre


8. CYP11B1 Mutations in patients with suspected Congenital Adrenal Hyperplasia (HAC) in the public neonatal screening in South Brazil: two case reports. Cristiane Kopacek, Federal University of Health Sciences of Porto Alegre (UFCSPA)


11. Aromatase deficiency caused by a Novel Mutation in the NADPH Cytochrome P450 Oxidoreductase. Amit V Pandey, Pediatric Endocrinology, University Children’s Hospital Bern


Category – Diagnostics


14. Why pediatricians need to know about the disorders of sex development: experience of 709 patients followed at a single specialized centre in 28 years. Mayra de Souza El Beck, Interdisciplinary Group for the Studies of the Disorders of Sex Development – School of Medical Sciences, State University of Campinas, Brazil

15. A Clinical and cytogenetic study of patients with Disorders of Sex Development (DSDs) Associated with Congenital Anomalies or Recognizable Syndromes. Mona El Gammal, Professor of Clinical Genetics and Head of the Human Genetics and Genome Research Division, National Research Centre
16. Clinical evaluations of different three patients with 46, XY disorder of sex development. **Betül Ersoy**, Celal Bayar University, Faculty of Medicine, Division of Pediatric Endocrinology, Manisa, Turkey

17. Combining clinical and genetic approaches in diagnosing a large Brazilian cohort of patients with 46,XY Differences of Sex Development. **Nathalia Lisboa Gomes**, Unidade de Endocrinologia do Desenvolvimento/ LIM42/SELA, Hospital das Clínicas. São Paulo, Brasil

18. Ovotesticular Disorder of Sexual Development due to 45X/46XY, inv dup(Yp)/46 XY: case report. **Guilherme Guaragna-Filho**, DSD Program (PADS), Hospital de Clínicas de Porto Alegre (HCPA) / Universidade Federal do Rio Grande do Sul (UFRGS), Porto Alegre, Brazil

19. When to investigate prostatic utricle in hypospadias? **Mila Torii Corrêa Leite**, Federal University of Sao Paulo

20. Profile of patients with disorders of sexual development in a reference hospital. **Rafael Miranda Lima**, Joana De Gusmao Children Hospital

21. The Utility Of AMH In The Prediction Of Testosterone Response To HCG Stimulation In Children With A Suspected DSD. **Angela K Lucas-Herald**, Developmental Endocrinology Research Group, University of Glasgow, Glasgow, UK

22. A clinical algorithm to diagnose differences of sex development. **Alejandra del Pilar Reyes**, Centre for Endocrinology and Metabolism, Hudson Institute of Medical Research, Melbourne, Victoria, Australia and Genetics Department, Hospital Infantil de México Federico Gómez, Mexico City, Mexico

23. Identification of an unbiased sample of persons with disorders (differences) of sex development in electronic medical records. **David E Sandberg**, Department of Pediatrics and Susan B. Meister Child Health Evaluation and Research (CHEAR) Center, University of Michigan Medical School

**Category – Genomics**

24. Integration of Next-Generation Mapping and Sequencing Technologies for Identification of Pathogenic Structural Variants. **Hayk Barseghyan**, Children’s National Medical Center

25. Low Frequency of Pathogenic Allelic Variants in the Disorders of Sex Development (DSD) Related Genes in Small for Gestational Age Children with Hypospadias. **Barbara Leitao Braga**, Unidade de Endocrinologia do Desenvolvimento/ LIM42/SELA, Hospital das Clínicas, Sao Paulo, Brazil

26. Challenges in identification, annotation, and interpretation of next-generation genome sequencing variants in DSD genes. **Emmanuèle Délot**, Center for Genetic Medicine Research/Children's National Medical Center

27. Successful virilization of a partial androgen insensitivity patient with a pathogenic missense variant in the ligand-binding domain of the androgen receptor with combined high-dose testosterone and aromatase inhibitor. **Sorahia Domenice**, Disciplina de Endocrinologia e Metabologia, Laboratório de Hormônios e Genética Molecular/LIM42, Hospital das Clínicas, Faculdade de Medicina da Universidade de São Paulo, São Paulo, Brazil

28. NR5A1 mutation screening disclose 11% of mutations in 46,XY DSD and male infertility. **Helena Fabbri-Scallet**, Center for Molecular Biology and Genetic Engineering - CBMEG, State University of Campinas, São Paulo, Brazil

29. Different clinical manifestations in three patients with the same pathogenic variant in the steroidogenic factor 1 gene. **Ochoa Molina Maria Fernanda**, Endocrinology Unit, Pediatric División, Pontificia Universidade Católica de Chile
30. Exomic sequencing uncovers novel genetic associations in children with hypospadias and neurodevelopmental abnormalities. Gabriella Gazdagh, Developmental Endocrinology Research Group, Royal Hospital For Children, University of Glasgow, Glasgow, UK

31. Importance of the number of metaphases analysed to exclude chromosomal DSD. Romina P. Grinspon, Centro de Investigaciones Endocrinológicas ‘Dr. Cesar Bergadá’ (CEDIE), CONICET-CEI-División de Endocrinología, Hospital de Niños R. Gutiérrez, Buenos Aires, Argentina

32. Whole Exome Sequencing Identifies Homozygous Missenses in INSL6 and WWOX Genes in Two Non-Related Infertile Men. Mara Sanches Guaragna, Human Molecular Genetics, Center for Molecular Biology and Genetic Engineering, State University of Campinas, Brazil

33. Use of a diagnostic DSD gene panel and implications for clinical care. Chloe Hanna, Department of Gynaecology, Royal Children’s Hospital, Melbourne, Australia

34. Cytogenetic spectrum of Ovotesticular Disorder of Sex Development in Egyptian DSD patients. Inas Mazen, Prof of Clinical Genetics & Endocrinology

35. Novel mutations in HSD17B3 in two cases of 46,XY Disorders of Sex Development. Taís Mazzola, Center for Investigation in Pediatrics, Faculty of Medical Sciences, UNICAMP

36. Development of testicular organoids to understand Disorders of sex development. Ken McElreavey, Human Developmental Genetics/Institut Pasteur

37. Mutations in CBX2 associated with gonadal anomalies in 46,XY and 46,XX individuals. Ken McElreavey, Human Developmental Genetics/Institut Pasteur


39. A rare case of apparently non-mosaic 47,XXY ovotesticular disorder of sex development: clinical, cytogenetic, molecular and immunohistochemistry features. Flávia Marcorin de Oliveira, Department of Medical Genetics and Genomic Medicine/School of Medical Sciences (FCM)/University of Campinas (Unicamp)

40. Sex Chromosome Mosaicism, two case reports. Karen Ramos Rodríguez, Child Health National Institute San Borja, Lima, Peru

41. SF1next–A Multicenter Study on Clinical and Genetic Effects of Human Steroidogenic Factor 1 Variants on Sex Development and Steroid Biology. Grit Sommer, Pediatric Endocrinology, Diabetology and Metabolism, Department of Pediatrics, Inselspital, Bern University Hospital, University of Bern, Switzerland

42. Molecular characterization of patients with 46,XY differences in sex development in a single tertiary center. Maria Sol Touzon, Hospital de Pediatría S.A.M.I.C. "Prof. Dr. Juan P. Garrahan"

43. NR5A1 Gene Mutation: Variable Phenotypes, New Variants, Different Outcomes. Małgorzata Wasniewska, Department of Human Pathology in Adult and Childhood, University of Messina, Messina, Italy

44. Context is the Key: A New Look at Dosage Sensitive Sex Locus Xp21.2. Ralf Werner, Department of Pediatrics and Adolescent Medicine, Division of Paediatric Endocrinology and Diabetes, University of Lübeck, Lübeck, Germany

45. An approach for identifying and evaluating Xp21.2 copy number variations in Patients with elusive aetiology of 46,XY gonadal dysgenesis. Ralf Werner, Department of Pediatrics and Adolescent Medicine, Division of Paediatric Endocrinology and Diabetes, University of Lübeck, Lübeck, Germany
46. Ovarian gonadoblastoma and dysgerminoma in two sisters with 46XY karyotype. Aysehan Akinci, Inonu University Medical Faculty Pediatric Endocrinology Department

47. Sexuality and Fertility Issues in a Large Cohort of 46,XY Disorders of Sex Development Individuals. Rafael Loch Batista, Developmental Endocrinology Unit, Hormone and Molecular Genetics Laboratory (LIM/42), Endocrinology Division, Internal Medicine Department, Medical School, University of São Paulo, Brazil

48. The Triple X syndrome impacts long-term health outcome negatively: a Danish nationwide registry study. Agnethe Berglund, Department of Molecular Medicine, Department of Endocrinology and Internal Medicine, Department of Clinical Genetics, Aarhus University Hospital, Denmark

49. Disorders of Sex Development associated with gender incongruence and emotional problems: a cross-sectional study. S.B. Cardoso, Hospital de Clínicas de Porto Alegre (UFRGS)

50. The Dutch DSD Together – a unique network. Hedi Claahsen-van der Grinten, Radboud DSD centre, Radboud University Nijmegen Medical Centre

51. Late-Diagnosed Ovotesticular disorder/difference of sex development: a case report. Eveline Gadelha Pereira Fontenele, Ambulatorio de Endocrinologia do Desenvolvimento e Gonadas/Hospital Universitário Walter Cantidio/Universidade Federal do Ceará

52. Challenges in management of late-diagnosed 46,XY disorder/difference of sex development: a case report. Eveline Gadelha Pereira Fontenele, Ambulatorio de Endocrinologia do Desenvolvimento e Gonadas/Hospital Universitário Walter Cantidio/Universidade Federal do Ceará

53. Transdermal estrogen therapy in 46,XY and 46,XX DSD patients with female identity – one clinical center experience. Aneta Gawlik, Medical University of Silesia, School of Medicine, Department of Pediatrics and Pediatric Endocrinology with the Subunit of Disorders of Sex Development, Katowice, Poland

54. Have surgical practices in the Intersex|DSD population changed in the last 2 decades? A 20-year picture of gonadectomies and genital surgical trends. Chloe Hanna, Department of Gynaecology, Royal Children’s Hospital, Melbourne, Victoria. Australia


57. The Evolution of DSD-Management at the University Children’s Hospital Zurich 1945-1970 - Learning points for future guidelines in a post-consensus era. Jürg C. Streuli, Center for Medical Humanities, University of Zurich, Switzerland AND University Children’s Hospital, Zurich, Switzerland

58. Differences of sex development with chromosomal mosaicism: characterization of histological findings and immunohistochemistry markers in gonads during childhood. Maria Sol Touzon, Hospital de Pediatría S.A.M.I.C. "Prof. Dr. Juan P. Garrahan"
Category – Psychosocial

59. Adolescent and adult patients in attendance by the HCPA – Gender disorder and construction of sexuality. C. A. Bridi Filho, PROTIG/HCPA – UFRGS

60. Parents’ participation in decision making on early genital surgery in patients with Disorders of Sex Development (DSD). Tatiana Prade Hemesath, Hospital de Clínicas de Porto Alegre

61. Understanding and communication around DSD according to the mothers and patients’ perspectives. Lia Mesquita Lousada Quintão, Unidade de Desenvolvimento do Serviço de Endocrinologia e Metabologia do Hospital das Clínicas da Universidade de São Paulo

62. Optimal Models of Delivering Care for Patients with DSD: Opinions of Different Stakeholders. David E Sandberg, Department of Pediatrics and Susan B. Meister Child Health Evaluation and Research (CHEAR) Center, University of Michigan Medical School

63. Timing of Reconstructive Surgery for Patients with Disorders/Differences of Sex Development (DSD): Preliminary Analysis of Stakeholder Views on Successful Outcomes. David E Sandberg, Department of Pediatrics and Susan B. Meister Child Health Evaluation and Research (CHEAR) Center, University of Michigan Medical School

64. Defining successful outcomes in people with differences / disorders of sex development: perspectives and priorities of different stakeholders. David E Sandberg, Department of Pediatrics and Susan B. Meister Child Health Evaluation and Research (CHEAR) Center, University of Michigan Medical School

65. DSD education for patients and parents: are principles alone adequate? David E Sandberg, Department of Pediatrics and Susan B. Meister Child Health Evaluation and Research (CHEAR) Center, University of Michigan Medical School

66. Decisional conflict in differences/disorders of sex development (DSD). David E Sandberg, Department of Pediatrics and Susan B. Meister Child Health Evaluation and Research (CHEAR) Center, University of Michigan Medical School

67. The holistic interplay of ethical and psychosocial perspectives in multidisciplinary DSD teams. Jürg C. Streuli, Multidisciplinary DSD-Working Group, University Children’s Hospital, Steinwiesstrasse 75, 8032 Zurich, Switzerland

68. Enhancing Professional Competence: Evaluation of the First Multidisciplinary Conference in Intersex for Frontline Professionals. Elaine Y.L. Tsui, Hong Kong Baptist University