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Ana Raquel Gouveia Freitas da Silva

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ABOUT ME

I am a recent medical specialist in Medical Genetics, with a greater focus in the last years in the area of prenatal diagnosis. I really like the clinical and laboratory part with the application of new NGS tools increasingly relevant nowadays. Not forgetting the inherent human, ethical and psychological aspects.
I enjoy reading a good book and I love summer and the sea.

WORK EXPERIENCE

01/12/2017 – 31/12/2017 Lisbon, Portugal
INTERNSHIP OF THE COMMON YEAR CHLC (LISBON CENTER) CHLC

01/01/2018 – 01/01/2023 Lisbon, Portugal
MEDICAL GENETICS SPECIALTY INTERNSHIP; INTERNAL PHYSICIAN WITH SPECIFIC TRAINING IN MEDICAL GENETICS, OBTAINING A SPECIALIST DEGREE (18,8 VALUES). CHLN (CENTRO HOSPITALAR DE LISBOA NORTE)

01/09/2023 – CURRENT Lisbon, Portugal
HOSPITAL ASSISTANT PHYSICIAN AT THE MEDICAL GENETICS SERVICE CHLN (CENTRO HOSPITALAR LISBOA NORTE)

Working in all areas of medical genetics (oncogenetics, cardiogenetics, neurogenetics, pediatrics) with a special focus on prenatal diagnosis and fetal malformations.

EDUCATION AND TRAINING

22/10/2010 – 20/06/2016 Lisbon, Portugal
INTEGRATED MASTER DEGREE IN MEDICINE (16,065 VALUES) NOVA Medical School, Faculty of Lisbon

12/10/2018 – 13/10/2018 Lisbon
POSTER - GOUVEIA SILVA R, MOLDOVAN O, MACHADO C, VITORINO E, CLODE N, SOUSA AB: "TYPE II GLUTARIC ACIDURIA – A RARE FORM OF PRESENTATION IN THE PRENATAL CONTEXT" Portuguese Association of Prenatal Diagnosis

14/10/2018 Porto, Portugal
28TH TRAINING COURSE OF THE COMPLEMENTARY MEDICAL GENETICS INTERNSHIP - PRE-IMPLANTATION GENETIC DIAGNOSIS (PGT) Department of Research and Postgraduate Support of São João Hospital

10/11/2018 Lisbon, Portugal
COURSE ON "BASIC GENETICS: ITS APPLICATION TO PRENATAL SETTING" Scientific Meeting of the Portuguese Association of Prenatal Diagnosis, Alfredo da Costa Maternity

15/11/2018 – 17/11/2018 Porto, Portugal
ORAL COMMUNICATIONS "COPY NUMBER VARIATIONS – SUSCEPTIBILITY LOCI ASSOCIATED WITH VARIABLE EXPRESSIVITY/ INCOMPLETE PENETRANCE – A RETROSPECTIVE STUDY" - GOUVEIA SILVA R, CUSTÓDIO S, SILVEIRA-SANTOS R ET AL Portuguese National Genetics´ Society Annual Meeting

08/04/2019 Lisbon
ORAL COMMUNICATION - SOARES M, GOUVEIA SILVA R, DAGGE A, CRUZ L, SANTO S, SOUSA AB: "MEDICAL INTERRUPTIONS OF PREGNANCY - 2018" Meeting of the Obstetrics and Gynecology Service, CHLN

05/10/2019 Viana do Castelo, Portugal
TRAINING WORKSHOP COURSE: "COMMUNICATION SKILLS IN DIFFICULT PRENATAL CLINICAL CONTEXTS" APDPN Scientific Meeting

26/08/2020 Almada, Portugal
ORAL COMMUNICATION - GOUVEIA SILVA R, MOLDOVAN O, COELHO H: "GENETIC TESTS - PRE-NATAL DIAGNOSIS" Garcia da Orta Hospital

30/10/2020 Lisbon
CLINICAL SESSION - GOUVEIA SILVA R: "ARTHROGRYPOSIS CONGENITA - SYSTEMATIC REVIEW" Genetics Department, CHLN

28/02/2021
VIRTUAL WEBINAR - FETAL MEDICINE FOUNDATION - "WEBINAR COURSE IN FETAL MEDICINE ON NOONAN SYNDROME"

28/03/2021
VIRTUAL WEBINAR - FETAL MEDICINE FOUNDATION - "WEBINAR COURSE IN FETAL MEDICINE ON NEUROSONOGRAPHY"

25/04/2021
VIRTUAL WEBINAR - FETAL MEDICINE FOUNDATION - "WEBINAR COURSE IN FETAL MEDICINE ON FETAL HYDROPS FETALIS"

25/05/2021
VIRTUAL WEBINAR - FETAL MEDICINE FOUNDATION - "WEBINAR COURSE IN FETAL MEDICINE ON FETAL GROWTH RESTRICTION"

20/06/2021
VIRTUAL WEBINAR - FETAL MEDICINE FOUNDATION - "WEBINAR COURSE IN FETAL MEDICINE ON CONGENITAL DIAPHRAGMATIC HERNIA"

04/2021 Lisbon, Portugal
PROTOCOL - GOUVEIA SILVA R, SOEIRO AND SÁ M, SOUSA AB: RECOMMENDATIONS FOR ACTION - NUCHAL TRANSLUCENCY (TN) > P99 (1ST TRIMESTER) Genetics Department, CHLN

01/10/2021 Aveiro, Portugal
FETAL CARDIOLOGY COURSE APDPN Scientific Meeting

01/10/2021 – 02/10/2021 Aveiro, Portugal
POSTER - GOUVEIA SILVA R, SOEIRO E SÁ M, LOUREIRO T, SOUSA AB: "FAMILY SYNPOLYDACTYLY AND THE FUTURE IN YOUR HANDS" APDPN Scientific Meeting

24/05/2021
ORAL COMMUNICATION - GOUVEIA SILVA R, MOLDOVAN O, SOUSA AB: "FETAL CEREBRAL VENTRICULOMEGALY - CLINICAL GUIDANCE" Obstetrics and Gynecology Service, CHLN

28/08/2021
EUROPEAN HUMAN GENETICS VIRTUAL CONFERENCE ESHG

10/03/2022
FETAL FIRESIDES CME LECTURE SERIES: MYELOMENINGOCELE (VIRTUAL) Continuing Medical Education Department, Children's Hospital of Philadelphia, USA

04/2022
PROTOCOL - GOUVEIA SILVA R, MOLDOVAN O, SOUSA AB: ACTION PROTOCOL - FETAL VENTRICULOMEGALY Genetics Department, CHLN

01/04/2022 – 30/04/2022 Barcelona, Spain
COURSE - IMPROVEMENT COURSE IN PRENATAL CLINICAL GENETICS Service of Maternal-Fetal Medicine at Hospital Clínic

23/04/2022 Barcelona, Spain
ORAL COMMUNICATION - GOUVEIA SILVA R: "IMPLEMENTATION OF EXOME SEQUENCING IN PRENATAL DIAGNOSIS AND IMPACT ON GENETIC COUNSELING: THE POLISH EXPERIENCE" Maternitat San Ramon, Barcelona

23/04/2022 Barcelona, Portugal
ORAL COMMUNICATION - GOUVEIA SILVA R: "WAARDENBURG SYNDROME TYPE 2 – PRENATAL PRESENTATION" Maternitat San Ramon, Barcelona (Multidisciplinary Meeting)

20/05/2022 – 21/05/2022 Lisbon, Portugal
PRE-CONGRESS COURSE: "A STEP BY STEP APPROACH TO COMMON AND CHALLENGING ULTRASOUND FINDINGS" UPDATES in Fetal Medicine, PT Meeting Center

27/05/2022 Coimbra, Portugal
ORAL COMMUNICATION - GOUVEIA SILVA R: "DISCORDANT ULTRASOUND FETAL SEX WITH NIPT, WITH REVIEW OF THE LITERATURE ON SEX DETERMINATION DISORDERS Maternity and Bissaya Barreto

11/04/2022 Lisbon, Portugal
CLINICAL SESSION - GOUVEIA SILVA R: "SINGLETON EXOME SEQUENCING OF 90 FETUSES WITH ULTRASOUND ANOMALIES REVEALING NOVEL DISEASE-CAUSING VARIANTS AND GENOTYPE-PHENOTYPE CORRELATIONS" (VIRTUAL FORMAT). Alfredo da Costa Maternity

27/05/2022 Coimbra, Portugal
CLINICAL SESSION - GOUVEIA SILVA R: "NIPT RESULT COMPATIBLE WITH TRISOMY 15 - DIAGNOSTIC HYPOTHESES" Bissaya Barreto Maternity

03/06/2022 Coimbra
"1ST FETOPLACENTAL PATHOLOGY COURSE OF THE PORTUGUESE SOCIETY OF PATHOLOGICAL ANATOMY" Centro Hospitalar Universitário de Coimbra

23/06/2022 Coimbra, Portugal
CLINICAL SESSION - GOUVEIA SILVA R: "MEGALENCEPHALY - REMEMBERING A CASE" Bissaya Barreto Maternity

15/07/2022 Lisbon, Portugal
CLINICAL SESSION - GOUVEIA SILVA R: "PROPOSAL FOR CLINICAL CRITERIA FOR EXOME IN PRE-NATAL" Medical Genetics Service, Hospital Santa Maria, CHULN

23/09/2022 Lisbon, Portugal
CLINICAL SESSION - GOUVEIA SILVA R: "REPRODUCTIVE GENETICS"; SELECTED CONFERENCE THEME: EUROPEAN HUMAN GENETICS CONFERENCE, VIENNA, AUSTRIA, 2022 Medical Genetics Service, Hospital Santa Maria, CHULN

09/2022
PROTOCOL - GOUVEIA SILVA R, MOLDOVAN O, DUPONT J, DIAS P, RODRIGUES M, SOEIRO AND SÁ M, TRAVESSA A, SOUSA AB: ACTION PROTOCOL - CLINICAL CRITERIA FOR EXOME IN PRE-NATAL MEDICAL Genetics Service, Hospital Santa Maria, CHULN

14/09/2022 – 17/09/2022 Barcelona, Spain
ORAL COMMUNICATION - GOUVEIA SILVA R, MOLDOVAN O, SOUSA AB: "A DIAGNOSIS THAT DOESN'T FIT?" - 32ND EUROPEAN MEETING ON DYSMORPHOLOGY

21/10/2022 Sesimbra, Portugal
COURSE - "NEUROSONOGRAPHY" APDPN Scientific Meeting

22/10/2022 Sesimbra, Portugal
POSTER - GOUVEIA SILVA R, CARVALHO I, FERREIRA L, PINHO M, COHEN A: "A TRIPLICATION INVOLVING THE 11P15 IMPRINTING REGION - A CLINICAL CASE" APDPN Scientific Meeting

22/10/2022 Sesimbra, Portugal
POSTER - GOUVEIA SILVA R, REBELO M, MOLDOVAN O, ARAÚJO A, (...) SOUSA AB: "EVALUATION OF PRENATAL DIAGNOSIS OF CONGENITAL HEART DISEASE IN A TERTIARY CENTER – 6-YEAR RETROSPECTIVE ANALYSIS" APDPN Scientific Meeting

04/11/2022 – 05/11/2022
9TH WORLD CONGRESS ON CONTROVERSIES IN PRECONCEPTION, PREIMPLANTATION AND PRENATAL GENETIC DIAGNOSIS (COGEN, VIRTUAL)

25/11/2022 – 26/11/2022 Lisbon, Portugal
POSTER - BORGES A, GOUVEIA SILVA R, SANTOS R, SOUSA AB, FERREIRA JC: "ARRAY-CGH VS CONVENTIONAL KARYOTYPE: THE POTENTIAL DIAGNOSTIC INCREASE IN THE PRENATAL CONTEXT" 6th National Congress of the Portuguese Society of Obstetrics and MMF

09/06/2023 – 13/06/2023
ORGANIZATION AND MODERATION OF A SYMPOSIUM AND A WORKSHOP INTEGRATED INTO THE ESHG 2023 ANNUAL CONGRESS, RESPECTIVELY: "DIAGNOSTICS IN PRENATAL SETTING - THE PRESENT AND THE FUTURE" AND "TERMINATION OF PREGNANCY: LEGISLATION AND ETHICS" ESHG Annual Conference, Glasgow 2023

27/10/2023 – 28/10/2023 Lisbon, Portugal
FETAL HEART COURSE Updates in Fetal Medicine

03/11/2023 – 04/11/2023
10TH WORLD CONGRESS ON CONTROVERSIES IN PRECONCEPTION, PREIMPLANTATION AND PRENATAL GENETIC DIAGNOSIS (COGEN, VIRTUAL)

08/12/2023 Paris, France
FETAL WINTER SCHOOL ERN ITHACA

Website https://ern-ithaca.eu/multidisciplinary-fetal-diagnostics-winter-school-2023-edition/

23/06/2024 – 27/06/2024 Lisbon, Portugal
FETAL MEDICINE FOUNDATION CONGRESSS

26/09/2024 – 27/09/2024 Lisboa
WORKSHOP: MEDICAL GENETICS AND PERSONALIZED MEDICINE - XI EDIÇÃO DO CONGRESSO NACIONAL DE ESTUDANTES DE MEDICINA (CNEM), ASSOCIAÇÃO NACIONAL DE ESTUDANTES DE MEDICINA (ANEM). FMUL, Hospital Santa Maria

LANGUAGE SKILLS

Mother tongue(s): **PORTUGUESE**

Other language(s):

	UNDERSTANDING		SPEAKING		WRITING
	Listening	Reading	Spoken production	Spoken interaction	
ENGLISH	C2	C2	C2	C2	C1

Levels: A1 and A2: Basic user; B1 and B2: Independent user; C1 and C2: Proficient user

PUBLICATIONS

2023

Not everything is always genetic - A prenatal presentation of tortuous and aneurysmal ductus arteriosus

Gouveia Silva R, Rodrigues M, Rebelo M: “Not everything is always genetic - A prenatal presentation of tortuous and aneurysmal ductus arteriosus”; Acta de Obstetrícia e Ginecologia Portuguesa; sem FI.

Abstract
Tortuous and aneurysmal ductus arteriosus (TADA) is a relatively rare condition, especially in the first and second trimesters. The ductus arteriosus has functional importance in the fetal circulation, being its aneurysmatic format characterized by saccular dilatation of the vessel. Majority of cases have a benign evolution, although they can be associated with syndromic forms and with important complications. We report the clinical management and echocardiographic follow-up in a newborn with this very early finding in prenatal setting.
Keywords: Aneurysmal ductus arteriosus; Prenatal setting; Early diagnosis.

Raquel Gouveia Silva, Márcia Rodrigues and Mónica Rebelo. Not everything is always genetic - A prenatal presentation of tortuous and aneurysmal ductus arteriosus. *Acta Obstet Ginecol Port.* 2023. Vol. 17(2):172-174. DOI:

Ref: AOGP-D-22-00050R1

Link https://www.euromedice.pt/pageflips/aogp_932765_abrjun_2023

2022

Prenatal phenotyping: A community effort to enhance the Human Phenotype Ontology

- Dhombres F, Morgan P, Chaudhari B, Filges I, Sparks T, Lapunzina P, Gouveia Silva R, (...) Robinson P. et al: “Prenatal phenotyping: A community effort to enhance the Human Phenotype Ontology”; 2022, doi: 10.1002/ajmg.c.31989; PMID: 35872606; Publicado no AJMG (American Journal of Medical Genetics), revista indexada com factor de impacto de 2,2.

Link https://doi.org/10.1002/ajmg.c.31989

2024

DYRK1A-related intellectual disability syndrome: a cohort of Portuguese patients

Introduction: DYRK1A heterozygous pathogenic variants have been shown to cause a syndromic form of intellectual disability (ID) with impaired speech development, features of autism spectrum disorder (ASD), microcephaly, and a recognizable facial gestalt that evolves with age. Patients can also present with gait disturbance or hypertonia, epilepsy, brain imaging, ocular, and foot anomalies. Methods: This is a cross-sectional study. Clinical data on patients with DYRK1A pathogenic variants identified at the Clinical Genetics Department of Santa Maria Hospital, in Lisbon, were retrospectively collected from medical records using a detailed clinical questionnaire. Results: We describe eight unrelated patients, six females and two males, aged 4 to 24. Fetal growth restriction (FGR) was present in 5/8, and microcephaly in 7/8. ID, ranging from mild to severe, and language impairment or absent speech were documented in all patients. ASD and/or stereotypic behavior were reported in 6/8. Five patients presented visual anomalies, most commonly optic disc pallor (in 4). Three main facial features were consistently reported: deep-set eyes, thin upper lip, and micro/retrognathia. Foot and hand anomalies were frequent. Discussion/Conclusions: Our cohort illustrates the variable degree of severity of a syndromic form of ID, which includes mild cases. Microcephaly and a typical neurobehavioral phenotype are in accordance with the literature, as well as some common dysmorphisms. Interestingly, optic disc pallor seems to be a frequent finding, highlighting the need for ophthalmological surveillance. Our study adds evidence to the existence of a consistent clinical phenotype of DYRK1A-related ID, hopefully contributing to increased awareness and improving the recognition of this entity.

R Gouveia Silva, J Rodrigues-Alves, J Dupont, O Moldovan, P Dias, M Rodrigues, A Sousa

2024

Termination of pregnancy for fetal malformations and severe genetic disorders: what are the laws in Europe?

Laws regarding termination of pregnancy for fetal anomalies (TOPFA) are extremely different, even among countries belonging to the European Union (EU) [1]. The EU Parliament recognizes women’s sexual and reproductive rights, including abortion rights, as fundamental, however it affirms member States’ autonomy to legislate on the issue, with huge variability regarding where and when TOPFA is allowed [2].
The information regarding abortion laws in European countries can be difficult to access for professionals working in prenatal genetics. In an attempt to fill this knowledge gap and discuss about the ethical implications of such a variable legislation within the EU, a workshop entitled “*Termination of pregnancy: Legislation and ethics*” was organized by the European Society of Human Genetics-Young (ESHG-Y) committee [3] within the context of the ESHG 2023 conference which took place in Glasgow In June 2023. The workshop was followed by ~120 in-person and 90 online participants.

Silvia Kalantari; Raquel Gouveia Silva; Mridul Johari; José Carlos Ferreira; Mirella Parachini

PROJECTS

01/09/2023 – CURRENT

Future type project for the CHLN Genetics Service - "Updating protocols for prenatal diagnosis"

The "type" project for the Genetics Service (Lisbon, CHLN) focusing on the area of prenatal diagnosis would aim to review the approach to congenital malformations and prenatal findings, in accordance with national, European, and international recommendations.

In addition to the protocols that I managed to carry out during my internship in Genetics, many areas of DPN remain to be covered in which we must review our approach, both obstetric and genetic. This is a constant need for the Service, being fundamental for the correct guidance and counseling for couples and pregnant women.
The protocols previously established in partnership with Obstetrics are limiting and dated 2016.

With technological advances and considering the increasing transition of arrayCGH studies to fetal exome, it is urgent to review the approach to various clinical contexts in prenatal care.
This project must consider:
Global scientific evidence (updated literature);
The approach was carried out in reference locations in DPN at national level (MAC, MBB), contacting those responsible for each center. This project can also be the starting point for creating action protocols at national level in the future.
The approach carried out in DPN centers abroad (Hospital Clinic, Necker Hospital, Great Ormond Street Hospital) contacting the prenatal teams at each center for this purpose and understanding the different ways of acting in an identical context.
The main objective of this project is to review the obstetric and genetic approach to DPN of various malformations/ ultrasound changes:
-Non-immune fetal hydrops
-Fetal growth restriction
-Omphalocele
-Posterior fossa defects
-Fetal nephrological pathology
-Minor markers
-Fetal skeletal pathology

Coordination with other DPN centers at national and international level will be relevant for the enrichment of our protocols; for the acquisition/updating of essential knowledge in daily clinical practice in prenatal diagnosis; to create links between services that allow the discussion of clinical cases in this area; also impacting the creation of training opportunities for interns undergoing specific training.
I have been working on the "Joint recommendations on delivering a fetal sequencing service" with the ISPD (Joint recommendations on delivering a fetal sequencing service) and the Policy and Ethics Committee EuroGentest Committee (ESHG) since September 2024.

30/06/2021 – 31/12/2022

Medical Internship Project - Assessment of prenatal diagnosis of congenital heart disease in a tertiary center – 6-year retrospective analysis

In accordance with the provisions of point 2.4.4. of the training program in the Medical Genetics specialization area, I planned and prepared a clinical research project in prenatal setting. The project was based on the activities of my Genetics´ department, mainly in the Prenatal Genetics consultation in collaboration with Pediatric Cardiology, specifically in congenital heart diseases. The first steps started from the following key question: "What factors condition the prognosis of Fallot Tetralogy, and should they be considered to inform the couple when deciding on prenatal care?", a question raised after a multidisciplinary meeting and which arose from the Prenatal Genetics consultation when counseling several couples, whose fetus has this heart disease. Many conditioning factors emerged for the practical execution of this project. Thus, a more comprehensive objective was defined, using the diagnoses of congenital heart defects from our hospital center (CHULN) in a prenatal context, with the Pediatric Cardiologist, Dr. Mónica Rebelo, providing invaluable help in collecting and analyzing this data. The project was entitled "Evaluation of prenatal diagnosis of congenital heart disease in a tertiary center – 6-year retrospective analysis". The choice of this project continued to consider my personal interest regarding the prenatal setting, specifically in relation to congenital heart diseases. On the other hand, the Genetics´ department was also interested in understanding whether our conduct has been the most appropriate in this area and in genetic counseling for couples, considering various factors, from the genetic diagnosis to the prognosis of the congenital heart disease itself.
This project's main objective was to identify cases of congenital heart disease diagnosed in a prenatal context from January 2016 to December 2021, at CHULN; analyze its relationship with some variables: gestational age when performing the fetal echocardiogram; pathological family history of heart disease; observe the types of congenital heart disease identified and categorize them; check which genetic diagnoses are most common; understand the correlation of heart disease with the existence or not of a genetic diagnosis; observe in which cases the pregnancy continued vs. medical interruption/fetal loss; verify whether the results of the fetal anatomopathological examination

corroborated the prenatal echocardiographic evaluation; observe whether the postnatal echocardiographic evaluation of cases that progressed and resulted in birth corroborated the changes observed prenatally.
The results of the project were presented in the form of a poster with discussion, at the APDPN annual scientific meeting, held in Sesimbra, in October 2022, at the Sesimbra Hotel; in the form of a poster, at the 26th SPGH Annual Meeting, in November 2022, at Convento São Francisco in Coimbra; and published in the department´s newsletter, in November 2022.

Link https://drive.google.com/file/d/1frbfY-MewLbg7CD3cOG7_Chpzb8RPE8P/view?usp=drive_link

NETWORKS AND MEMBERSHIPS

Affiliations

- Partner of SPGH (Portuguese Society of Human Genetics) since 2018 to date.
- Member of the European Society of Human Genetics (ESHG) since 2020 to date.
- Member of the Portuguese Association of Prenatal Diagnosis since 2021 to date.
- Member of the "ESHG-Young" Commission (Vice-President; 2021-2025)
- ERN-ITHACA network - vice-president representing Hospital Santa Maria and genetics´ department, Lisbon, Portugal
- Member of the European Working Group in prenatal diagnosis (ERN-ITHACA): WG Fetal Medicine (start: January 2023)
- Member of the ESHG Education Committee (2022-2025).

Collaborations

- Collaborator of the National Registry of Congenital Anomalies (RENAC) of the Ricardo Jorge Institute (case records from 2019 to the present).
- HPO (Human Phenotype Ontology) project (2022), through the improvement of descriptions and characterizations of certain phenotypes observed in the prenatal context, namely in the cardiac and skeletal areas, including some specific terms of venous duct agenesis and the "salt and pepper" pattern of prenatal renal ultrasound.
- Involvement in the Unique, Understanding Chromosome and Gene Disorders project, as an ESHG-Young member, through the recruitment of volunteers to translate pamphlets on pathologies already on the Unique website, as well as through proposals to create new pamphlets for other relevant pathologies for patients and their families (2021 - to date).
- Scientific Committee Participation at the Annual European Meeting on Dysmorphology with Prof. Koen Devriendt and Prof. Alain Verloes.

HONOURS AND AWARDS

14/02/2020

"Intellectual Development Disorder associated with the MED13L gene - National Case Series" – CHLN, Pediatrics Journey

Prize for 2nd best oral communication; Gouveia Silva R, Moldovan O, Afonso J, Sousa S, Soares C, Soares G, Dias P, Antunes D, Amorim M, Lopes F, Maciel P, Venâncio M, Sá J, Sousa AB: "Intellectual Development Disorder associated with MED13L gene - National Case Series"