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MESTRADO
ACONSELHAMENTO GENÉTICO

Relatório de Estágio

Rodrigo Martínez Martín

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2022

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Rodrigo Martínez Martin Relatório de Estágio

M.ICBAS **2022**

INSTITUTO DE **CIÊNCIAS BIOMÉDICAS ABEL SALAZAR**

Relatório de Estágio de 2º ano

Relatório de Estágio de Candidatura ao grau
de Mestre em Aconselhamento Genético
submetido ao Instituto de Ciencias Biomedicas
de Abel Salazar da Universidade do Porto.

Orientador – Dra. Isabel Lorda Sánchez

Categoría – Médica/ Chefe associada de Serviço.

Hospital Fundación Jiménez Díaz.

Coorientador – Dra. Carolina Lemos

Categoria – Professora Catedrática

Instituto de Ciencias Biomédicas Abel Salazar

Index

Introduction

This “relatorio” was written by Rodrigo Martínez Martín, 2nd year student of the master's degree in Genetic Counselling at the Instituto de Ciências Biomédicas Abel Salazar (ICBAS), a centre belonging to the University of Porto. This “relatorio” aims to summarise the academic activities carried out by the student during his professional practical year.

The student carried out the practical activity corresponding to his professional year in the Genetics Service of the Fundación Jiménez Díaz Hospital. Hospital belonging to the Autonomous Community of Madrid (Spain). This activity began on 13 September 2021 and ended on 12 June 2022 (9 months), with a total of more than 1240 hours, a sufficient number of hours, according with the regulations for the satisfactory completion of this training activity.

The course was carried out under the supervision and tutoring of Dr. Isabel Lorda Sánchez, medical geneticist and associate head of the Genetics Service of the aforementioned hospital.

The student carried out a research project in the field of genetic counselling under the title: "Parental views, motivations and attitudes towards the genetic testing in Spain. Influential factors".

Academic curriculum

University of La Laguna Tenerife (Spain) Sept. 2008 – Jan. 2015	Degree in Biology (specializations: Health Biology and Molecular and Cellular Biology)
CESIF Madrid (Spain) Nov.2015 – Sept. 2016	Master's Degree in International Sales and Marketing Management of Pharmaceutical and Allied Companies
University of Murcia Murcia (Spain) Nov. 2019 – April. 2020	University Expert in Medical Genetics and Genomics
University of Porto Porto (Portugal) Oct. 2020 – currently	Master's Degree in Genetic Counselling

Professional curriculum

Applied Genomics/ Hops. La Candelaria Tenerife (Spain) Sept. 2012 – Jan. 2013	Research Assistant
Swansea University Swansea (United Kingdom) April 2014 – Dic. 2014	Research Assistant
MSD/ L'Oréal Madrid (Spain) June 2016 – Sept. 2017	Marketing
Angelini/ Roche Madrid (Spain) Oct. 2017 – Oct. 2020	Sales Representative

Estágio profissionalizante

The professional “estágio” is part of the second year of the master's degree in genetic counselling and is compulsory to obtain the master's degree, which qualifies the student to practice the profession.

The second academic year of the master's degree in Genetic Counselling at the Universidade do Porto/ ICBAS is made up of the professional “estágio” (50 ECTS) and a research seminar (10 ECTS) whose purpose is to create, develop and execute a research project related to genetic counselling, the outcome of which will be the publication of the results in a scientific journal.

Objetives

1. Establish relationship and clarify clients’ concerns and expectations

    1.1 Establishes an environment which facilitates client to expression of feelings, anxieties, beliefs, and expectations and considers clients’ experiences.
    1.2 Identifies client needs
    1.3 Enables clients to make informed choices about the implications of their family history.
    1.4 Takes appropriate action to meet identified needs with the agreement of the client.

2. Make appropriate and accurate genetic risk assessment.

    2.1 Ascertains sufficient medical, family and personal information from the client to make appropriate genetic risk assessment.
    2.2 Ascertains medical information from other sources to confirm family information and diagnosis.
    2.3 Understands the patterns of inheritance and the underlying mechanisms by which genetic disease may occur.

3. a. Convey clinical and genetic information to clients, appropriate to their individual needs. Genetic Counselling

3. b. Explain options available to the client, including the risks, benefits and limitations. Genetic Counselling

3. c. Evaluate the understanding of the individual related to the topics being discussed. Genetic Counselling

3. d. Acknowledge the implications of individual and family experiences, beliefs, values and culture for the genetic counselling process.

    3.1 Provides information about the genetic disorder appropriate to the client’s assessed needs, reflecting their values, religious and cultural beliefs and preferences.
    3.2 Provides information based upon appropriate interpretation of genetic and clinical knowledge.
    3.3. Communicates with respect to the genetic risk assessment and possible options.
    3.4 Supports dissemination of information about the genetic disorder to at risk relatives by the client.

4. Make an assessment of clients' needs and resources and provide support, ensuring referral to other agencies as appropriate.

4.1 Ascertains psychological needs of the individual or family.
4.2 Respecting clients' preferences, provides support and makes referrals to other agencies (such as psychologist or patient support groups).
4.3 Identify and support clients' access to local, regional and national resources and services.

5. Use of a range of counselling skills to facilitate clients' adjustment and decision-making.

5.1 Uses safe, effective and appropriate counselling skills to support clients to make adjustments and decisions.

6. Document information including case notes and correspondence in an appropriate manner. Clinical Psychology

6.1 Uses a systematic approach to collecting and maintaining comprehensive and accurate records that detail placement the rationale underpinning any interventions.
6.2 Maintains confidentiality and security of written and verbal information.

7. Find and utilise relevant medical and genetic information for use in genetic counselling.

7.1 Collects, evaluates and uses relevant information about the genetic disorder in question.
7.2 Critically appraises current evidence to inform practice and professional development.
7.3 Disseminates evidence of good practice and service improvement through verbal and written media.

8. Demonstrate knowledge of sequencing technologies, challenges and limitations of Genomics in clinical practice and practical skills for analysis and variant interpretation

8.1 Uses knowledge on Next generation sequencing, whole exome and whole genome analysis while discussing with patients the testing options.
8.2 Understands the analytical and clinical sensitivity and specificity of these tests.
8.3. Is aware of the potential application of emerging technologies development of Genomic services in their country and Europe.
8.4. Understands the theory of sequence analysis and the use of genome analysis tools and bioinformatics databases.
8.5 Understands and applies the different Guidelines and Standards for the Interpretation, classification and Report of Sequence Variants.
8.6 Critically appraises the limitations and challenges faced by current sequencing technologies and additional, incidental and secondary findings from whole genome sequencing.

9. Demonstrate ability to organise and prioritise a case load.

9.1 Addresses client needs in a sensitive and fair manner making best use of resources available
9.2 Prioritises according to patient need.

10. Plan, organise and deliver professional and public education

10.1 Facilitates understanding of how genetics impacts on affected individuals, their families, partners and carers.

10.2 Seeks to raise awareness of available services and resources related to genetic healthcare.
10.3 Acts as a resource for other professionals and lay groups.

11. Establish effective working relationships to function within a multi-disciplinary team and as part of the wider health and social care network.

11.1 Promotes patient-centred care in partnership with the client, their family, and appropriate care providers.
11.2 Facilitates communication via a strong multidisciplinary network of professional and lay colleagues.
11.3 As appropriate, co-ordinates patient and family care.

12. Contribute to the development and organisation of genetic services.

12.1 Evaluates own practice and that of others in the light of new evidence and modifies practice appropriately.
12.2 Uses skills of critical appraisal to consider how new evidence may contribute to the improvement of service organisation and delivery.
12.3 Actively seeks opportunities to meet with colleagues to discuss professional issues and innovations in care, in order to disseminate best practice and improve standards of care.
12.4 Actively seeks opportunities to collaborate with colleagues in audit and research that has the ultimate aim of improving client care

13. Practice in accordance with an appropriate code of ethical conduct.

13.1 Upholds professional standards of safe and ethical practice at all times.
13.2 Uses professional standards of practice to evaluate own and others' performance.
13.3 Recognises the duty to seek professional advice if standards of care are threatened.
13.4 Contributes to the debate on ethical challenges in genetic practice.
13.5 In normal circumstances discloses information about individuals to appropriate third parties only with the client's permission.

14. Recognise and maintain professional boundaries and limitations of own practice.

14.1 Recognises practice limitations and demonstrates referrals to other health professionals when appropriate.
14.2 Consults other health professionals when the client's needs fall outside the scope of genetic practice.
14.3 Refers clients to colleagues when necessary.

15. Demonstrate reflective skills and personal awareness for the safety of individuals and families.

15.1 Demonstrates reflective practice, which informs future clinical interactions.
15.2 Maintains a portfolio recording reflection on practice.
15.3 Accesses counselling and/or clinical supervision to underpin and enhance practice.

16. Present opportunities for clients to participate in research projects in a manner that facilitates informed choice.

16.1 Enables clients to make an informed choice on whether to participate in a research project or not.

17. Demonstrate continuing professional development as an individual practitioner and for the development of the profession.

17.1 Actively seeks opportunities to update knowledge and skills and reflects on the implications of these for own practice and that of professional colleagues.

Institution

The professional training course in genetic counselling was carried out at the Genetics Service of the Fundación Jiménez Díaz Hospital.

The Genetics Service of the Hospital Fundación Jiménez Díaz was founded in 1962, being the first hospital with a genetics service in Madrid. It is made up of more than 30 professionals with different profiles, including physicians, biologists, biochemists, pharmacists, pre-doctoral and post-doctoral researchers, laboratory technicians and administrative staff.

The Service is organized into three sections: Clinical Genetics Consultation, Cytogenetics and Molecular Genetics. It also has several multidisciplinary functional units:

- Reproductive Advice Unit (with the Obstetrics and Gynecology Service).
- Preimplantation Genetic Diagnosis Unit (with the Reproduction Unit).
- Hereditary Cancer Unit (Participation in the Hereditary Cancer Programme of the Community of Madrid).
- Neurogenetics Unit (diagnosis and predictive diagnosis and follow-up).
- Neurosensorial Unit.

Facilities

- Outpatient consultations: this area has a waiting room, a secretary's office and four offices where general postnatal consultations, reproduction and prenatal consultations and monographic consultations are carried out.
- Molecular Genetics laboratory area (I and II): where acid nucleic extraction, pre- and post-PCR room, electrophoresis room, arrays, capillary sequencing, and mass sequencing are carried out.
- Cytogenetics laboratory area: with cell culture area and sterile chamber, optical and fluorescence microscopy room. Image analysis and automatic karyotyping.
- Preimplantation Diagnosis and Non-invasive Prenatal Diagnosis Area: with sterile chamber and fluorescence microscopy room.
- Research and Teaching Area: where the biobank and a multipurpose area are located.
- Genomics Area (to support the Instituto de Investigación Sanitaria - Fundación Jiménez Díaz or IIS-FJD).

The laboratory is UNE EN ISO 9001 certified. It also participates as a European and Spanish representative of Quality Control programmes: AMQN, GenQA, AEGH, EUROGENTEST and AEDP.

The Genetics Service of the Fundación Jiménez Díaz Hospital carries out its activities in the areas of care, teaching, research, and management.

Care activity

The Service offers a care portfolio that includes Genetic Counselling, Genetic Diagnosis and Preconception Diagnosis, Prenatal (invasive and in maternal blood) and Postnatal, as well as Pharmacogenetic studies and Therapy in rare diseases.

The Genetics Service collaborates with different services from different specialities to set up multidisciplinary units for the diagnosis and clinical follow-up of possible genetic diseases. It also forms, together with the Obstetrics and Gynecology Service, two transversal units: the Prenatal Diagnosis Unit and the Assisted Reproduction Unit.

Its healthcare work is spread over four hospitals in the Community of Madrid, with the Fundación Jiménez Díaz University Hospital being the hospital where the preparation and analysis of samples are referred. As an example of activity in terms of care within these hospitals in 2019, the following number of consultations were carried out in the four hospitals:

Hospital Fundación Jiménez Díaz	
First medical appointment	2.923
Subsequent appointments	2.249
Total	**5.172**
Hospital Rey Juan Carlos	
First medical appointment	661
Subsequent appointments	356
Total	**1.017**
Hospital Universitario Infanta Elena	
First medical appointment	769
Subsequent appointments	421
Total	**1.190**
Hospital General de Villalba	
First medical appointment	308
Subsequent appointments	151
Total	**459**

Patient Circuit

In a global manner and summarizing, the passage of a patient through the hospital's genetics service would be as follows:

1.- Referral by the specialist doctor to the Genetics Service

In order for the patient to have access to the genetics service in a public (and not private) manner, he/she/them must be referred by a specialist doctor and cannot be referred by a family doctor.

2.- Obtaining an appointment

Generally, the patient obtains an appointment for the genetics service through a digital platform, either that of the Community of Madrid or that of the Quirón Salud group (the

company that manages the hospital). In other cases, if the patient does not have this platform, the appointment can be made by post or telephone.

3.- First medical consultation

The patient enters the waiting room located at the entrance to the genetics service where they can be seen by the secretary to confirm their attendance and appointment. It is also possible for the patient to carry out this procedure telematically via the Quirón salud application or via the machines located at the entrance to the hospital and/or the Genetics Service. Subsequently, the patient is called for the genetic appointment. Prior to the consultation, the genetics doctor has had access to the patient's history and has studied the case and the reason for the consultation, so that the genetics doctor has been able to prepare for the consultation. During the first medical consultation, the geneticist has the opportunity to examine the patient if necessary, to collect personal and family history, as well as to inform about the details of the specific test and disease, its reliability, extent and possible results to be obtained, and the expected time delay until the report is obtained. During this consultation, the geneticist will resolve any doubts the patient may have. It should be noted that the delay times until the results are obtained are particularly short in this Genetic Service, being, for example, in the case of using the NGS technique, generally between 2 and 3 months.

4.- Obtaining the sample

Once the geneticist has considered what type of genetic test is suitable for the case (if necessary), the patient will go to the place where the sample (usually blood) is taken. Generally, the sample is taken on the same day as the first consultation, but this may vary according to the patient's wishes.

5.- Presentation of the case at the Service Meeting

Generally, this step does not occur, but if the case is particularly complex, the case is presented at the department meeting, which takes place weekly. This step can take place before or after the sample collection, depending on the geneticist's assessment.

6.- Last consultation: delivery of results

In this consultation, the patient is informed of the results and the clinical implications of the results, if any, are explained. A genetic report is given to the patient, indicating the validity of the results. During this consultation, the medical geneticist will resolve any doubts that the patient may have regarding themselves or their relatives, always respecting data protection. In addition, the medical geneticist may refer the patient to different medical specialties depending on the results obtained.

Teaching activity

- Participation in training programmes at the Faculty of Medicine and the School of Nursing at the Autonomous University of Madrid, among others.
- Direction of undergraduate and master's degree final projects.
- Training of undergraduate students and predoctoral and postdoctoral researchers and laboratory technicians.
- Specialized health training for residents in other specialties of Medicine, Biology, Pharmacy and Chemistry.

Research activity

The service is part of one of the 27 research groups of the Instituto de Investigación Sanitaria Fundación Jiménez Díaz within the Area of Genetics and Genomics, Group of Genetics and Genomics of Rare and Complex Diseases. It also collaborates with basic and clinical groups, both national and international.

Human Resources

Head of Unit	Dra. Carmen Ayuso García (Doctor)
Associated Heads of the Unit	Dra. Mª José Trujillo Tiebas (Biologist) Dra. Isabel Lorda Sánchez (Doctor) Dra. Marta Rodríguez de Alba Freiría (Biologist)
Doctors	Dr. Saud Tahsin Swafiri (Doctor) Dra. Fiona Blanco Kelly (Doctor) Dra. Fermina López Grondona (Doctor) Dra. Ana Isabel Sánchez Barbero (Doctor)
Research/Molecular Diagnostics Staff	Dra. Almudena Ávila Fernández (Biologist) Dra. Ana Bustamante Aragonés (Biologist) Dra. Rosa Riveiro Álvarez (Biologist) Dra. Carolina Sánchez Jimeno (Biologist) Dra. Inmaculada Martín Mérida (Biologist) Dra. Berta Almoguera Castillo (Biologist)
Laboratory technicians	Dña. Rocío de Libertad Cardero Merlo D. Jesús Gallego Merlo Dña. Inés García Vara Dña. Ascensión Jiménez Pardo Dña. Laura Horcajada Burgos D. Fernando Infantes Barbero D. Miguel Ángel López Martínez D. Camilo Vélez Monsalve
Quality Manager	Dña. Ruth Fernández Sánchez
Auxiliary Nursing Care Technician	Dña. Amelia Cambronero Crespo
Secretary	Dña. Aurora Marín Escrich Dña. Teresa Ciudad
Research team	Dra. Marta Cortón Pérez (Biologist) Dr. Pablo Minguez Paniagua (Biologist) Dra. Irene Perea Romero (Biologist) +++

Care activity

The student spent the entire internship period at the same center, the Hospital Universitario Fundación Jiménez Díaz; however, during the first three months he spent the afternoons learning and developing skills in the Molecular Diagnostics laboratory. There he had the opportunity to continue learning about the analysis of genetic sequences using the NGS technique. The student worked in the genetics department from September 2021 to June 2022 (10 months) adapting to the hospital's own timetable, which are morning hours (from 8:00 am to 15:00 pm). In addition, as mentioned above, the student developed complementary activities in the afternoon (from 15:00 pm to 18:00 pm) during the first months of his stay.

During his activity in the Molecular Diagnostics laboratory, the student was reviewing cases of patients in whom the possible causal mutation of their disease had not yet been found and he had the opportunity to solve some of these cases. In particular, the student learned the importance of using specific gene filters in each case in order to improve the efficiency of the search for the causal gene and to preserve the patient's own privacy by preventing unexpected findings that have nothing to do with the patient's pathology.

In addition, the student had the opportunity to participate in various activities, among which the following stand out:

- Medical consultations (as an assistant or with active participation).
- Participation and attendance at meetings of the genetics service and with other services (neuropaediatrics, ophthalmology, etc.).
- Review of clinical cases
- Preparation of the consultation
- Preparation of different reminder and clarification documents for families/people attending Preimplantation Genetic Diagnosis consultations.
- Participation in the presentation of clinical cases
- Communication of results to patients.
- To deepen the need, or rather the possible non-necessity, of genetic testing for people who wish/need to undergo a change of gender. Looking for scientific evidence, etc
- Participation and attendance at congresses and courses related to clinical genetics.
- Preparation and development of the research project: "Parental views, motivations and attitudes towards the genetic testing in Spain. Influential factors".

The assistance activity carried out by the student in the genetic consultations was coordinated by his tutor Dr. Isabel Lorda Sánchez. Generally, the student was in a different consultation with a different medical geneticist each day of the week. In this way, the student was able to learn about the different medical profiles and ways of working with the different doctors. The student played a progressively more active role in consultations during his stay in the genetics department, moving from a purely observational role to do/performance genetic counselling consultations with patients, the availability of which depended on the complexity of the case and the work rhythm of the department.

All the doctors with whom the student consulted are prepared to see any case in any specialty; however, each of them specializes in different fields:

- Dr. Isabel Lorda Sánchez: Preimplantation Genetic Diagnosis.
- Dr. Fiona Blanco Kelly: Ophthalmology
- Dr. Saud Tahsin Swafiri: General
- Dr. Fermina López Grondona: Paediatrics
- Dr. Ana Isabel Sánchez Barbero: Ophthalmology and Paediatrics

Within the organization of the Genetics Service, most of the days the doctors had organized the days by specialty. The priority was always given to Pre Natal-Diagnosis cases. The student was free to choose which doctor he would be with that day, and even, if another doctor had a complex case or one of special interest, he could be in that specific consultation.

Data

The student had the opportunity to attend **1019 consultations**. The student made a record of all of them (in annex) taking into consideration the following parameters: date, patient number (deleted from the attached file for data privacy reasons), age, gender, medical geneticist, type of visit, specialty, reason for the visit, symptoms, findings (if any), diagnosed or suspected disease, altered gene (confirmed or suspected), type of inheritance, incidental finding, observed general psychological state of the patient, observed nervousness of the patient at the entrance of the consultation (if applicable), observed nervousness of the patient at the exit of the consultation (if applicable) and other comments. It should be noted that in the patient's observed state of nervousness, a record was made if and when conditions allowed, i.e. when the patient's condition was apparently clear. In addition, this recording was a specific exercise of the student, it is likely that it did not correspond to the reality of the moment, it is a purely subjective assessment.

A series of data defining the activity carried out during the consultations attended and participated in by the student is specified below:

1.- Number of patients seen with each genetics doctor:

Doctor	Number of patients
Dra. Isabel Lorda Sánchez	319
Dra. Fiona Blanco Kelly	213
Dr. Saud Tahsin Swafiri	4
Dra. Fermina López Grondona	281
Dra. Ana Isabel Sánchez Barbero	202
Total	**1019**

2.- Number of patients seen corresponding to each medical speciality:

Medical speciality	Nº patients	Percentage
Cancer	170	16,7%
Cardiology	43	4,2%
Dermatology	5	0,5%
Digestive	3	0,3%
Endocrinilogy	34	3,3%
Fertility	47	4,6%
General Genetic	44	4,3%
Hematology	4	0,4%
Nephrology	31	3,0%
Neurology	106	10,4%
Ophtalmology	106	10,4%
Otorhinolaringology	18	1,8%
Pneumology	3	0,3%
Transexuality	6	0,6%
PND	77	7,5%

	Speciality	Nº Patients	Percentage
Pediatrics		215	21,1%
	Dermatology	2	0,9%
	Endocrinology	11	5,1%
	General Genetic	16	7,5%
	Hematology	1	0,5%
	Nephrology	7	3,3%
	Neurology	164	76,6%
	Ophtalmology	12	5,6%
	Otorhinolaringology	1	0,5%
PGD		109	10,7%
	Cancer	14	13,0%
	Cardiology	11	10,2%
	Dermatology	2	1,9%
	Endocrinology	2	1,9%
	General Genetic	43	39,8%
	Hematology	1	0,9%
	Nephrology	4	3,7%
	Neurology	17	15,7%
	Ophtalmology	14	13,0%

As can be seen in the table, the type of patients most frequently seen by the student in consultation correspond to the speciality of "paediatrics", i.e. children (215 children; 21.1% of the patients seen). Of these, the majority (76.6%) come from the speciality of neurology, with the reason for the consultation in most of them being Autism Spectrum Disorder (ASD) and development disorders. For this type of consultation, a detailed anthropometric examination and measurement becomes essential, with a focus on facial features, sometimes by simple observation with the naked eye and sometimes by using measuring tools. These examinations,

together with the family history, are essential in guiding the diagnosis and in making decisions about which type of genetic test the medical geneticists should order. It is not surprising that paediatrics is the most frequent speciality due to the large number of congenital and/or early onset genetic diseases.

The second most frequent type of patient comes from cancer (170, 16.7%). In this type of consultation, it becomes more important to collect family data, with special emphasis on gender, age, relationship and type of primary cancer. It is highly recommended to ask for the medical inform of the family history of cancer to corroborate what the patient tells us. It is common to confuse primary cancers that occurred a long time ago, especially ovarian cancer. In this type of consultation, we generally find two "types" of test: diagnostic, which in the case of the student represented 67% of the cases, and predictive, with 33% of the consultations carried out. In terms of gender, the balance was 76% female and 24% male in the cases reported. The mean age was 52 years for all cases, 57 years for diagnostic cases and 43 years for predictive cases.

The third most frequent type of consultation was PGD. All these consultations were carried out under the direction of Dr. Isabel Lorda Sánchez. The Genetics Service of the Hospital Fundación Jiménez Díaz is a reference center for this type of consultation in the entire Community of Madrid. Patients and couples with diverse genetic alterations come to Dr. Isabel Lorda's appointment from all over the Community of Madrid because they wish to have healthy children without a specific genetic disorder (chromosomal or specific gene alterations). The process of getting to the genetic consultation becomes quite tedious and uninformative for these couples. It has been a comfort to observe these couples leaving the consultation with most of their questions answered and with the confidence that they are in the right place. Most of the questions received by the patients in these types of questions have to do with timing and not so much about the technical procedure. It is common to observe surprised reactions from patients when they hear about the different possibilities they have to be sure that they will have a free pathology-free children, as well as the probability of success of the process (around 20% for most cases). This is a type of consultation in which a huge diversity of feelings converges. Clarifying doubts and maintaining a facilitating role is key for these patients/partners. Generally, patients will come to the consultation twice: the first time for genetic counselling and for them to decide whether or not to continue with the process and the second time, called the 1st informative consultation (which can become the 2nd informative consultation) in which patients bring their relatives (normally parents and/or siblings, with priority given to carriers of the genetic alteration) to carry out a genetic test on them with the aim of better "mapping" the characteristic markers of the altered allele/chromosome and carry out the selection of the embryo with absolute certainty. It is important to add that all cases that undergo PGD must be approved by the National Commission for Assisted Human Reproduction in order to continue with the process. This commission ensures that all PGD procedures carried out in Spain are ethical and justified.

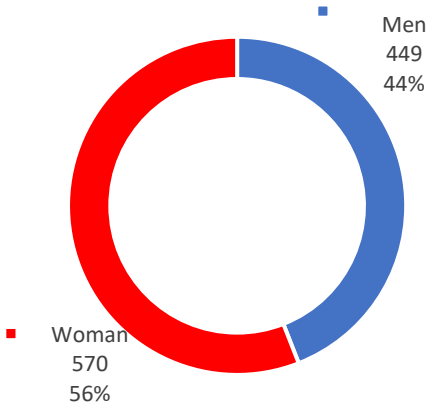
Following PGD consultations, we find genetic consultations derived from Neurology and Ophthalmology. It is not surprising that Ophthalmology is among the pathologies most reported by the student, as the Fundación Jiménez Díaz is one of the reference centres in this speciality at the genetic level. Its research role in this area stands out. The management of ophthalmology patients in the genetics department of the Fundación Jiménez Díaz Hospital is a perfect example of how research can be of great use in clinical practice and vice versa. The student had the opportunity to communicate research results to patients with a clinical diagnosis made years

ago. It was very gratifying for the student to partly observe the relief of the patients when they received confirmation of the molecular results and to hear their gratitude that their search for the reason for their disease was over. There is growing hope in these patients for treatment with gene therapy. Few patients are candidates for these treatments, but this is certainly that this is only the beginning of such treatments. Regarding the neurology referrals, the student had the opportunity to attend and participate in 106 consultations, where he was able to learn about numerous pathologies. The student was particularly struck by the Huntington's predictive consultations. These consultations are attended by patients suspected of being carriers, in some cases affected because they seem to manifest some subtle symptoms of Huntington's disease. These patients must undergo a psychological check-up before the genetic test can be carried out, the aim of this check-up being to verify that the patients are in good psychological condition to obtain the result. The student observed a multitude of attitudes in these patients, ranging from apparent calmness and lack of interest to fear and restlessness. In addition, the student also observed different mechanisms used by the patients to handle such a difficult situation, some rely on luck, others cling to the percentages (50%) so that, for example, if they have two siblings who are affected, the patient thinks that there is no way that he/she can be a carrier and, therefore, future affection, etc.

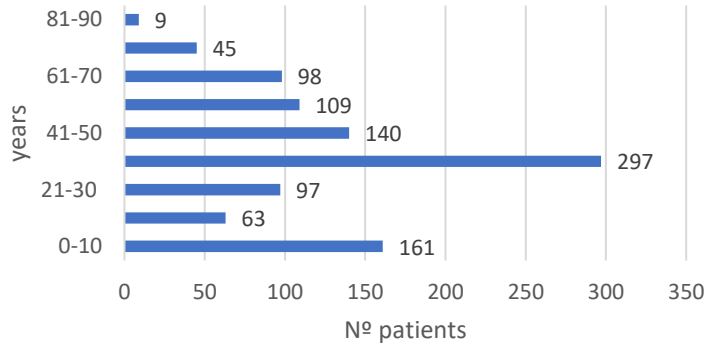
Finally, the student finds it interesting to highlight two other consultations: the PND and that of patients coming from endocrinology whose reason for consultation is to be transsexual. Regarding PND consultations, it is worth highlighting the hospital's prioritisation of this type of consultation and its results. In these consultations, time plays an important factor in the decision-making process of the parents. These consultations have a very important psychological character, patients often enter the consultation with high emotional stress, fears, and uncertainty. It is vital to be empathetic but calm, to answer all questions clearly and to tell them how long it will take to obtain the possible results. The diagnostic tests available to us are invasive tests (amniocentesis from 14 weeks of gestation and chorionic villus sampling from 11-12 weeks of gestation) and non-invasive tests (free fetal DNA in maternal blood). About consultations with transsexuals, the student wonders about the need for genetic testing of transsexuals. Especially when there is a disparity between the recommendations for carrying out the test or not in the guidelines followed by the different autonomous communities in Spain, which means that in some autonomous communities these people are obliged to take the test and in others they are not. It is surprising that in the recent bibliography on the subject there are studies that show no significance with respect to genetic findings in transsexuals and non-transsexuals and that in the Community of Madrid a genetic test was a mandatory requirement to be able to continue with the process of gender reassignment.

From a more general point of view, the following graphs show that 44% were men and 66% were women. The most frequent age range of the patients seen was between 31 and 40 years old. The average age of the patients seen was 37 years.

Patient's Gender:

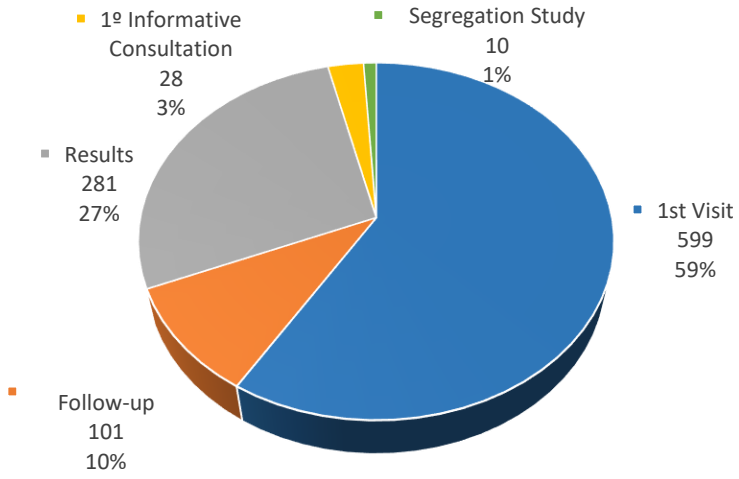


Patient's Age:



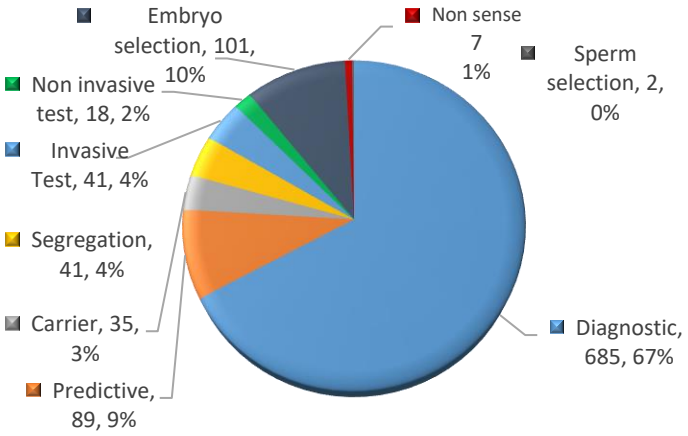
Regarding the type of consultation in terms of the time of consultation, we find:

The majority of the patients registered by the student (59%) were first-time visitors to the genetics consultation of that hospital. 27% of the registered patients were coming for a results delivery consultation (more on this later). 10% of the patients came for a follow-up consultation.



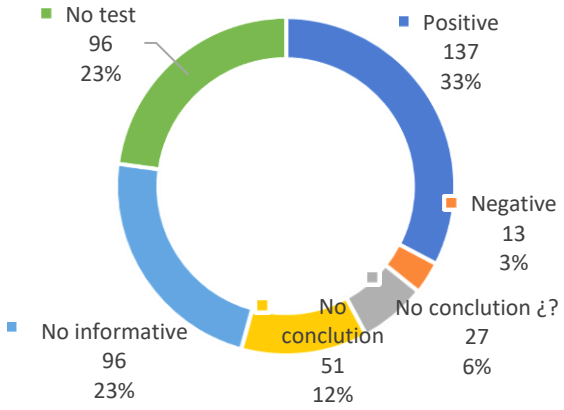
This generally occurs when the patient was not a candidate for the genetic test in the first consultation and in this new consultation it is possible that he/she would be a candidate, also because he/she is again referred by the specialist under suspicion of another genetic disease or because the results of the previous test were inconclusive and it is possible that now a re-analysis of the test performed can be carried out so that, thanks to the new reclassified variants, he/she can have a molecular diagnosis. With 3% we have the queries of 1st informative consultation corresponding to the PGD process. With 1% we have the segregation study consultations that occur when a genetic alteration has been detected in a family member whose significance is uncertain, but which could be the cause of the disease and the study is carried out in family members (generally close in terms of kinship), to help reclassify the variant found.

Regarding the objective and/or type of test to be performed or carried out on the patient, the student attended different types of cases:



Most of them were intended for diagnosis in non-pregnant affected patients (67%). Tests performed on parents for embryo selection under the PGD process accounted for 10%. Predictive tests accounted for 9% and carrier tests, etc.

Regarding the results obtained for the patients whose results consultation the student had the opportunity to attend:



We found that 33% of them were positive or conclusive, i.e. the genetic cause of the disease was found. Surprisingly, in 23% of the cases the genetic test was not carried out because it was not indicated for that case. In this type of case, the figure of the medical geneticist is fundamental, as he/she is the person who, according to his/her criteria and following established guidelines, decides when the genetic test is indicated. 23% of the results were not informative, i.e. the cause of the disease was not found, but this does not mean that the cause is not genetic. 12% of inconclusive results, i.e. some variable or genetic alteration was found that could be the cause of the disease but this has not yet been clarified. 6% of results that the student has taken the liberty of reclassifying as "No conclusion ¿?", which are results without conclusion but which, due to the variables/alterations found, make clarification even more complicated, such as finding two causal alterations in a patient that cause two similar diseases of incomplete penetrance in different genes.

Scientific meetings

The student had the opportunity to attend all genetics service meetings, which were held weekly and lasted 1h30min - 2h. In these meetings, particularly complicated cases were presented, either because of the complexity of their clinical or molecular diagnosis or because of their complexity in terms of the approach to the patient's consultation. The student had the opportunity to present a case on early onset bilateral hearing loss.

In addition to these meetings, the student had the opportunity to attend different meetings with other medical specialties, including meetings with ophthalmology (once a week) and neuropediatric (once every 15 days). These meetings were particularly enriching for the student, as they provided him with another point of view on how to approach the same patient.

Work carried out in the field of genetic counselling

The student produced an informative document for PGD consultations, explaining the processes to be followed, the basic concepts and answering the most frequently asked questions.

Contributions of the training programme

The student considers that his stay in this service was vital for the acquisition of medical knowledge about genetic pathologies and the diversity of genetic causes. The student also had the opportunity to learn a great deal of technical language, especially about the description of phenotypic manifestations.

The student had the opportunity to practice and expand their knowledge of risk calculation, interpretation of results and reporting.

The student tested and developed their communication and empathy skills. Especially useful for the genetic counsellor. He learned not to take certain future reactions of the patient for granted and to maintain a neutral stance at difficult moments for the patient.

The experience in the service helped him to evaluate the facts more and better and not so much the conclusions, to be able to think more critically and to question under the prism of evidence-based reasoning.

He learned to use the main sources of information on genetic pathologies and their variants with skill.

Improved their knowledge of the search for and classification of causal variants.

Final considerations

The humanization of genetics.

Although it is true that genetics, and specifically human genetics, is a field yet to be explored and advanced, countless discoveries and advances have been made in the field of clinical genetics. Clinical genetics has taken on a more than considerable size in terms of content and usefulness for it to become a specialty in Spain, as is the case in the rest of Europe. Linked to this progress and knowledge, the participation of the rest of the population is necessary. Projects such as IMPaCT are a clear example of this. In a context in which citizen participation is necessary and in which informed consent is ethically and legally indispensable, the transfer of knowledge to the population about what genetics is and what it can do for them is fundamental and basic. Are we completely sure that when patients sign the informed consent form, they know what they are really validating, or do they believe that it is just another procedure and only know the possible benefits and how slightly their samples will be treated? Therefore, due to the increasing knowledge, the universalization of human genetics and patient autonomy, this discipline needs professionals to ensure that patients fully understand all aspects of the discipline. In this educational process, science and emotions are mixed. Genetic counsellors are professionals specifically trained for this purpose, so it is necessary that this figure becomes a regular professional in genetic services and not a difficult figure to find in them. The existence of a genetic counsellor should be a component in validating the competitiveness of a genetic service. Just as there are species that act as verifiers of ecosystem health (so-called biomarkers), a genetic counsellor should be a marker whose existence or not speaks to the service's commitment to the correct understanding and psychological well-being of the patient.

Scientific activity

Participation in conferences, seminars and courses

1.- X Jornada de Cardiogenética, Hospital Clínico Universitario Virgen de Arrixaca. 26th november 2021

2.- Programa Y.O.D.A. (Young, Old, Development, Advance) de mentoring del Colegio Oficial de Biólogos de la Comunidad de Madrid. January – september 2022

3.- 17ª Jornada de Investigación Traslacional y Medicina de Precisión: Medicina de Precisión, más allá del genoma. 3rd february 2022

4.- VIII Jornada Interdisciplinar sobre avances em enfermidades raras, IBIMA – RARE. 4th march 2022

5.- Programa "Genomic-Oriented Medicine", International Precision Medicine Forum. Instituto de Investigación Sanitaria Valdecilla. 28 de febrero al 11th march 2022

6.- Jornada sobre enfermedades raras neurológicas de la Comunidad de Madrid, Red RAREGenomics. 17th march 2022

Collaboration with patient associations

The student collaborated actively in the patients' association "Dravet Foundation Spain" as part of the team involved in the human genome programme whose aim is to carry out molecular diagnosis of patients with symptoms like those described for Dravet Syndrome and who, for some reason, have not had access to these genetic tests. The most frequent cause being that their hospital does not carry out these tests. To do this, the student analyzed the results obtained from using NGS techniques on their samples, carrying out a prior filtering of target genes and later analyzing the resulting genes in search of alterations that explain their pathology.

Research project

The student's research was that of the research project "Parental views, motivations and attitudes towards the genetic testing in Spain. Influencing factors". Its objectives were to explore the attitudes, motivations, and views of Spanish parents towards genetic testing for any suspected genetic disease but taking into account psychosocial factors that may influence such as ethnicity, socioeconomic status, educational level, age and previous knowledge of genetic testing. It also aims to better understand the influence of early genetic counselling.

The study is in the process of being written up and published in a scientific journal.

Arrixaca
Hospital Clínico Universitario
Virgen de la Arrixaca

CERTIFICADO DE ASISTENCIA

Don/Dña. Rodrigo Martinez Martin

Ha asistido a la

X Jornada de Cardiogenética

Celebrado el 26 de noviembre de 2021. Actividad de Formación Continuada (12 horas).

ERN GUARD-HEART

Dr. Juan R. Gimeno Blanes
Unidad CSUR-ERN de Cardiopatías Familiares

Arrixaca CSUR. Cardiopatías familiares · SOCIEDAD MURCIANA DE CARDIOLOGÍA · cibercv ciberer · IMIB Instituto Murciano de Investigación Biosanitaria · Servicio Murciano de Salud · D-Corazón A.F.A.C.M.S. · SEPAF Sociedad Española de Patología Forense

Colegio Oficial de la Psicología de Madrid

CERTIFICADO

Que **RODRIGO MARTINEZ MARTÍN** ha participado como **TUTELADO** en el **PROGRAMA Y.O.D.A. (Young, Old, Development, Advance) DE MENTORING DEL COLEGIO OFICIAL DE BIÓLOGOS DE LA COMUNIDAD DE MADRID**, desde el mes de enero de 2022 hasta el mes de septiembre 2022

De acuerdo a la **METODOLOGÍA MINERVA**, del **COLEGIO OFICIAL DE LA PSICOLOGÍA DE MADRID** (*)

Y para que así conste y surta los efectos oportunos, firman el presente certificado en Madrid, a 1 de octubre 2022.

UNIÓN INTERPROFESIONAL

Unión Interprofesional Comunidad de Madrid
ASOCIACIÓN DE COLEGIOS PROFESIONALES

Colegio Oficial de Biólogos de la Comunidad de Madrid

Colegio Oficial de la Psicología de Madrid

El Secretario General	*El Secretario*	*La Secretaria*
D. Jaime Cabrero García	*D. Juan E. Jiménez Pinillos*	*Dª. Nuria Mateos de la Calle*

(*) De acuerdo con el Convenio específico de colaboración entre el Colegio Oficial de la Psicología de Madrid, el Colegio Oficial de Biólogos de la Comunidad de Madrid y la Unión Interprofesional de la Comunidad de Madrid para el desarrollo de la metodología de mentoring en el ámbito de los Colegios Profesionales y en virtud de lo expuesto en la estipulación segunda de dicho Convenio.

Hospital Universitario Fundación Jiménez Díaz
Grupo quirónsalud

IIS FJD
INSTITUTO DE INVESTIGACIÓN SANITARIA
FUNDACIÓN JIMÉNEZ DÍAZ

CERTIFICADO

D./Dña.Rodrigo Martinez Martin

Ha obtenido la acreditación correspondiente a la

17 ª Jornada sobre Investigación Traslacional y Medicina de Precisión: Medicina de Precisión, más allá del genoma

Celebrada el 3 de febrero de 2022

Esta actividad docente, con nº de expediente 07-AFOC-04206.1/2021 y realizada de forma online, ha sido acreditada por la Comisión de Formación Continuada de las Profesiones Sanitarias de la Comunidad de Madrid-Sistema Nacional de Salud, con **0,5 créditos** de formación continuada para las profesiones: Bioquímica (esp. Sanitaria), Enfermería, Farmacia, Medicina, Psicología clínica, Química (esp. Sanitaria). Enseñanza no reglada y sin carácter oficial.
Los créditos de esta actividad formativa no son aplicables a los profesionales, que participen en la misma, y que estén formándose como especialistas en Ciencias de la Salud.

Madrid, 3 de Febrero de 2022

Carmen Ayuso
Directora Científica
Instituto de Investigación Sanitaria
Fundación Jiménez Díaz

Consuelo Martín de Dios
Directora Gerente
Fundación Instituto Roche

Fundación InstitutoRoche

Comisión de Formación Continuada
SISTEMA NACIONAL DE SALUD
Comisión de Formación Continuada de las Profesiones Sanitarias de la Comunidad de Madrid

Av. Reyes Católicos, 2 Madrid-28040

VIII JORNADA INTERDISCIPLINAR SOBRE AVANCES EN ENFERMEDADES RARAS IBIMA-RARE

Crea conciencia, hazte visible

MÁLAGA, 4 DE MARZO DE 2022

Certifica que el/la Sr./Sra.:

Rodrigo Martínez Martín

Ha asistido a las **VII Jornada Interdisciplinar sobre Avances en Enfermedades Raras IBIMA-RARE**. Crea conciencia, hazte visible, celebrada en formato híbrido (presencial y en streaming), en Málaga el día 4 de marzo de 2022.

Málaga, 4 de marzo de 2022

Miguel Ángel Medina
Coordinación de IBIMA-Rare

Raquel Yahyaoui
Coordinación de IBIMA-Rare

RODRIGO MARTÍNEZ con D.N.I. Nº: 08891628N

ha participado en el programa

"GENOMIC-ORIENTED MEDICINE"

del International Precision Medicine Forum

celebrado en formato online del **28 febrero al 11 de marzo de 2022** con una duración de **16 horas lectivas,** habiendo superado todos los requisitos necesarios para recibir este Diploma de Acreditación.

Y para que conste, se expide el presente certificado en Santander

Dr. José Luis Fernández Luna
Coordinador de Genética
HUMV

Dr. Francisco Galo Peralta Fernández
Director de Gestión
IDIVAL

Madrid, 17 de marzo de 2022

CERTIFICADO DE ASISTENCIA

Red RAREGenomics. Jornada sobre enfermedades raras neurológicas de la Comunidad de Madrid

Celebrada el jueves 17 de marzo de 2022 de 10:00 a 13:00 h en el Hospital Universitario Fundación Jiménez Díaz

D. Rodrigo Martínez Martín

ha asistido a la actividad docente en la fecha arriba mencionada.

Dra. Carmen Ayuso
Directora del IIS-FJD, UAM
Jefa del Dpto. de Genética Clínica
Hospital Universitario Fundación Jiménez Díaz

MASTER DEGREE IN GENETIC COUNSELLING – MAG (ICBAS, UP)

PROFESSIONALIZING INTERNSHIP

2nd year periodical assessments and specific clinical or laboratorial rotations

– SUPERVISORS' ASSESSMENT FORM –

Student: RODRIGO MARTINS MARTIN

Clinical rotation: CLINICAL GENETICS

Service/Laboratory: GENETICA

Institution: IIS-HU FUNDACION JIMENEZ DIAZ

Training period: from 01/09/2021 to 30/06/2022

Local tutor (name and position): Dr Tona Blanco Kelly, Consultant
Dr Isabel Lorda Sanchez Consultant

Global Evaluation

(Please circle one: 1 - Fail; 2 - Pass; 3 - Good: 4 - Very Good; 5 – Excellent; and fill-in global score)

Professionalism and punctuality	Engagement and participation	Interpersonal relationships	Scientific knowledge	Global score: (0-20)
1 2 (3) 4 5	1 2 3 (4) 5	1 2 3 (4) 5	1 2 (3) 4 5	14

Comments:
The student was attentive and showed interest, but sets his own roadmaps
He has been punctual, applied and sociable, but he asked few questions and easily assumed ~~has~~ to have knowledge of a pathology just by having been in consultation.
It was difficult to guide him because he had established in advance what he considered to be interesting or important

Date 28/10/2022

Supervisor's signature

Tona Blanco Kelly MD, PhD

MASTER DEGREE IN GENETIC COUNSELLING – MAG (ICBAS, UP)

PROFESSIONALIZING INTERNSHIP

2nd year periodical assessments and specific clinical or laboratorial rotations

– SUPERVISORS' ASSESSMENT FORM –

Student: RODRIGO MARTINEZ MARTIN

Clinical rotation: ALL AREAS (PRENATAL, REPRODUCTIVE, CANCER ...)

Service/Laboratory: GENETIC DEPARTMENT

Institution: FUNDACION JIMENEZ DIAZ

Training period: from __/09/2022 to __/06/2022

Local tutor (name and position): DR. ISABEL LORDA
ASSOCIATED CHEF

Global Evaluation

(Please circle one: 1 - Fail; 2 - Pass; 3 - Good: 4 - Very Good; 5 – Excellent; and fill-in global score)

Professionalism and punctuality	Engagement and participation	Interpersonal relationships	Scientific knowledge	Global score: (0-20)
1 2 (3) 4 5	1 2 3 (4) 5	1 2 3 (4) 5	1 2 (3) 4 5	14

Comments:
He has shown interest during his attendance at the consultations but fundamentally in psychological issues rather than in the genetic bases of illness
Good relationship with patients and relatives

Date 27/10/2022

Supervisor's signature