FUNDACIÓN RAMÓN ARECES

The vision of the patients
Genomic Medicine

entidad de utilidad pública
feder
FEDERACIÓN ESPAÑOLA DE ENFERMEDADES RARAS

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Index

- What is FEDER?
- State of the art:
  - Diagnosis
  - Treatmet
  - A global challenge
- Why a Genomic Strategy?
- Patient Engagement

What is FEDER?

What is FEDER?

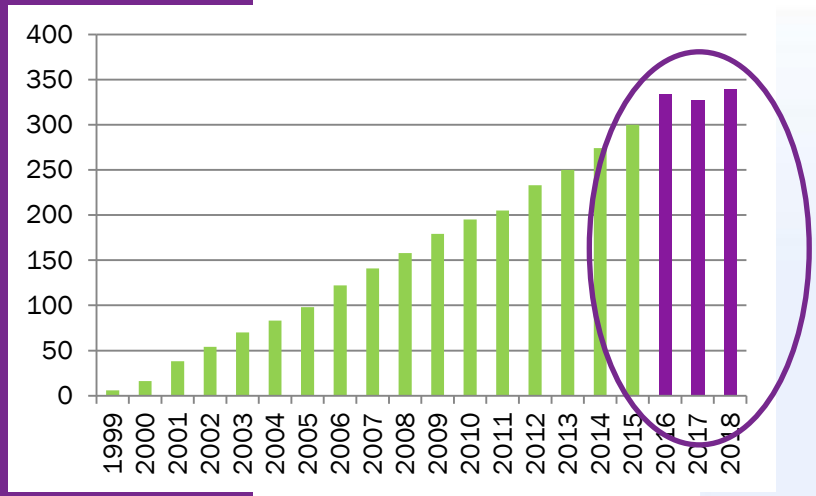
- National alliance of patient organization.
- Founded in 1999
- 358 member patient organisations
- Outreach to over 95.000 patient
- 50+ staff, offices in Andalucía, Cataluña, Comunidad Valenciana, Extremadura, Madrid, Murcia, País Vasco.
- 2 Million € Budget

Membership data

59% → National

41% → Autonomous Community

1.282 diseases



ALIBER
ALIANZA IBEROAMERICANA DE ENFERMEDADES RARAS

EURORDIS
RARE DISEASES EUROPE

RARE DISEASES INTERNATIONAL
A EURORDIS INITIATIVE

What is FEDER?

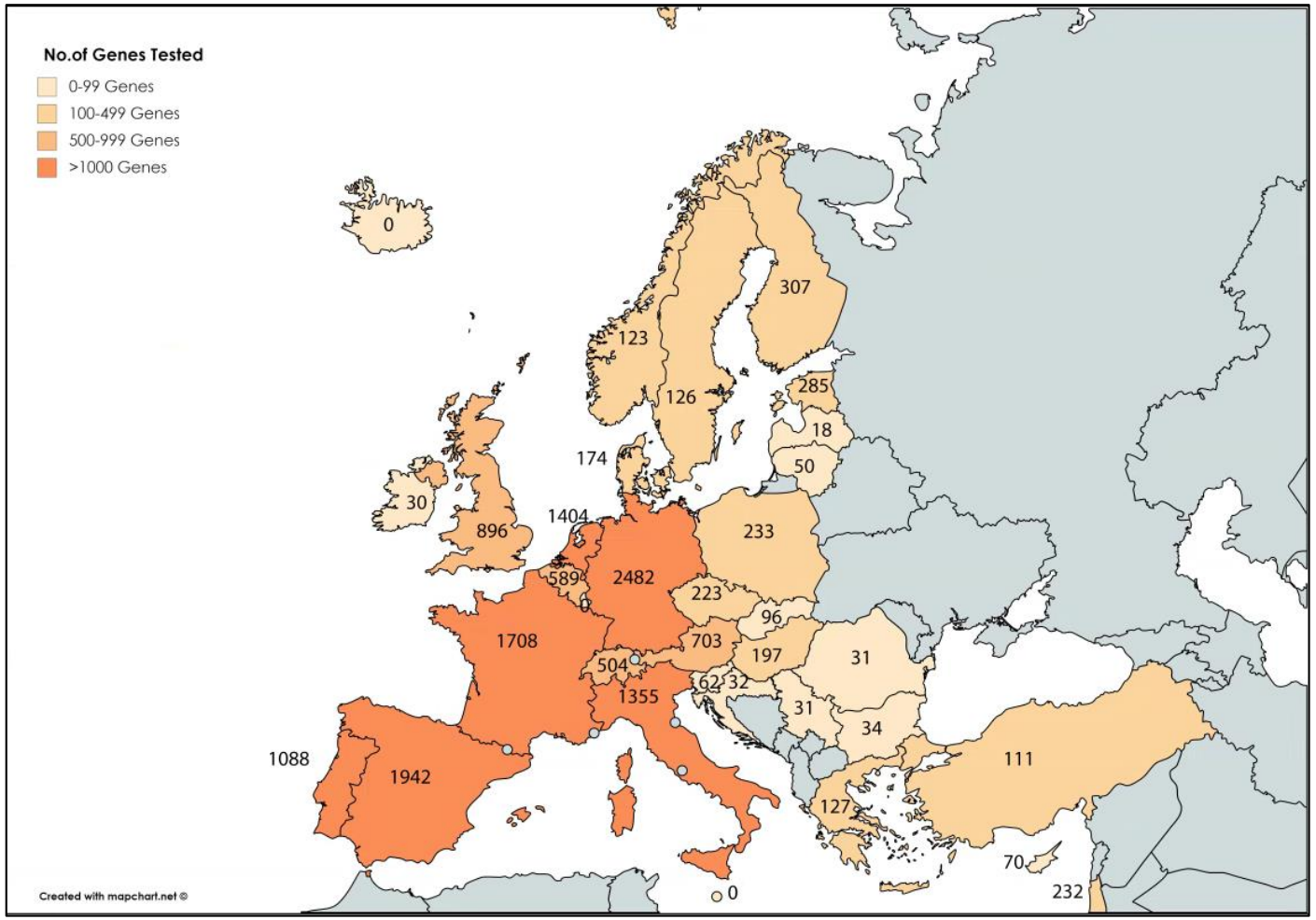
PEOPLE	MEMBERS	SOCIETY
		
Information and Orientation Service	Associative Movement Consultancy Service	Political incidence and social mobilisation
Psychological Attention Service	Associative Movement Training	Awareness Campaigns
Legal Aid and Educational Attention	Associative Movement Grants	Divulgation activities
Health products Access Programme	Associative Participation	Channels 2.0
Undiagnosed cases orientation and Advisory Committee	Communication Service for Associative Movement	Inclusion
Professionals'training		Investigation and knowledge

State of the Art

Diagnosis: Europe

State of the art

Gene Tested (Source SoA Orphanet)



Diagnosis: Europe - Spain

Genetics as a Medical Specialty

Eur J Hum Genet. 2017 Dec; 25(Suppl 2): S53–S59

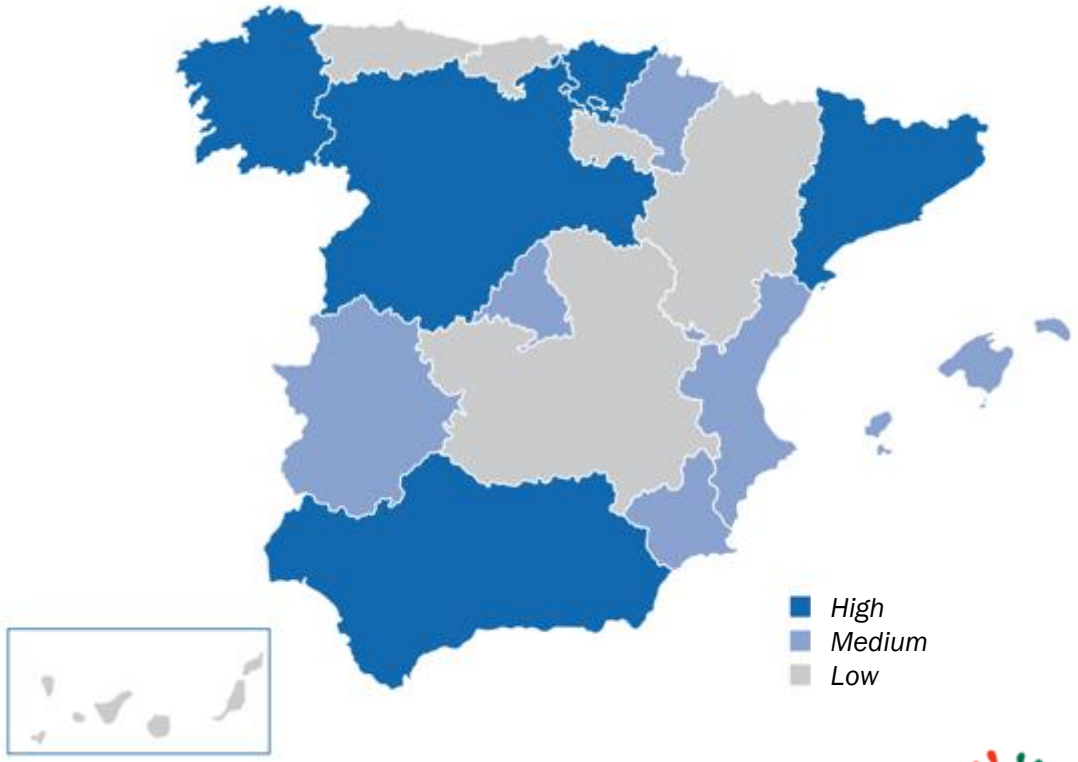
Country	Year/birth	Training yrs
Austria	2006	6
Belgium	2017	6
Bulgaria	2006	4
Croatia	2017	5
Czech Rep.	2009	4
Denmark	1996	5
Estonia	2009	4
Finland	1981	
France	1995	4
Germany	1992	5
Greece	2018	
Hungary	2012	4
Iceland	2015	5
Ireland	2012	4
Italy	1970	4
Latvia	2000	5
Lithuania	2004	
Malta	2005	5
Netherlands	1987	4
Norway	1973	4
Poland	2003	5
Portugal	2001	5
Romania	2008	4
Slovakia	2006	4
Slovenia	2001	5
Spain	2014	4
Sweden	1992	
UK	1984	4

State of the art

Personalized and Precision Genomic Medicine: CC.AA

State of the art

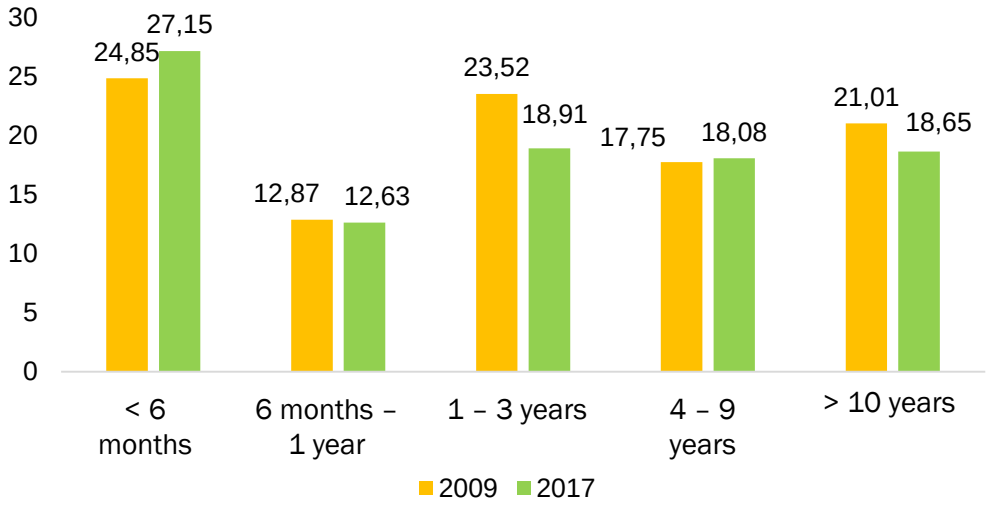
Implementation development



State of the art

Diagnosis: Spain

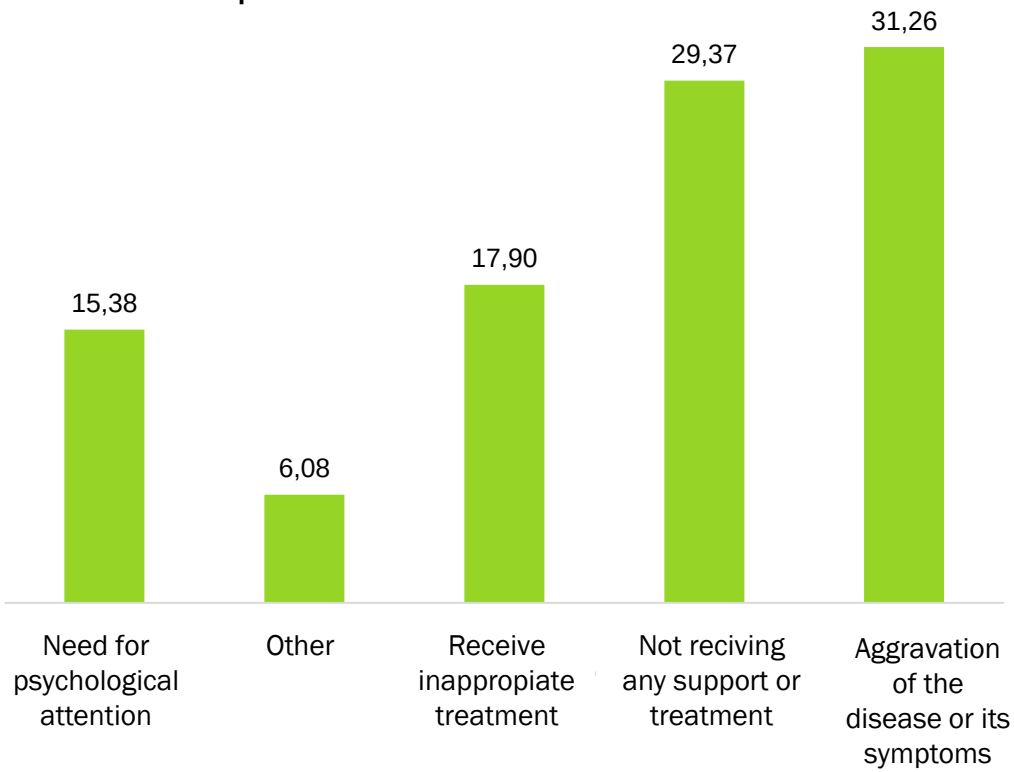
- 49.68% of them has gotten a delayed diagnosis.
- Almost one of five people with RD (18.65%) have taken more than 10 years to get an accurate diagnosis, and around 18.08%, from 4 to 9 years.
- 39.78% had a diagnosis before the first year of life (including 1.84% who had a prenatal diagnosis).



Study on the situation of Social &Health Needs of Spanish People with Rare Diseases

Diagnosis: Spain

- 31.26% of the respondents claimed that having a delayed diagnosis had worsened their disease or its symptoms; 29.37% argues that this delay meant neither receiving any backing nor treatment; and the 17.90% points out that this delay led to receive an inadequate treatment.



Study on the situation of Social &Health Needs of Spanish People with Rare Diseases

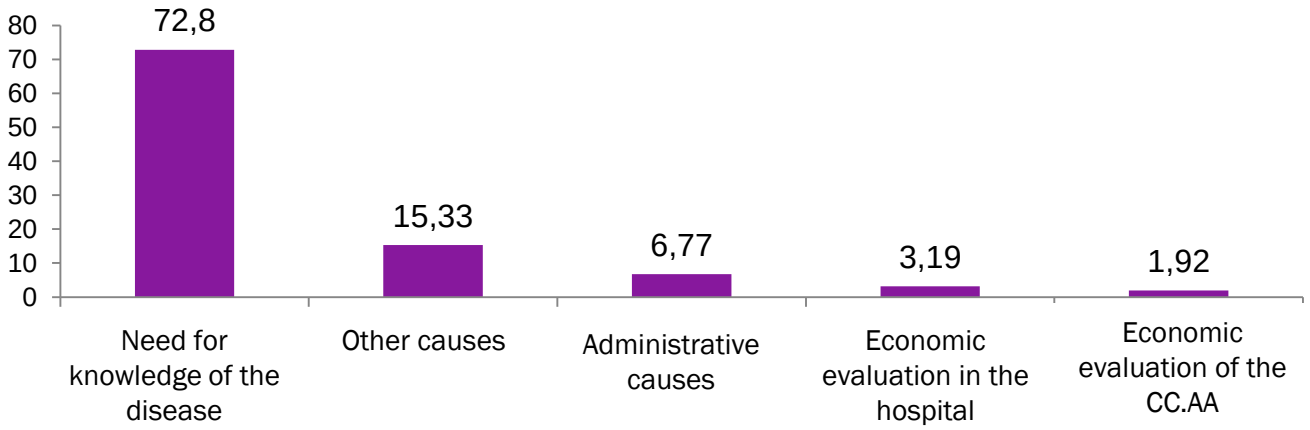
State of the art

State of the art

Diagnosis: Spain

- Regarding the causes of delay in diagnosis, in **72.8% of the cases, ignoring the disease is the main reason**.

- Other causes are related to administrations (6.77%), the economic evaluation in hospitals (3.19%) and, in little cases, the economic evaluation in the Autonomous Community (1.92%).



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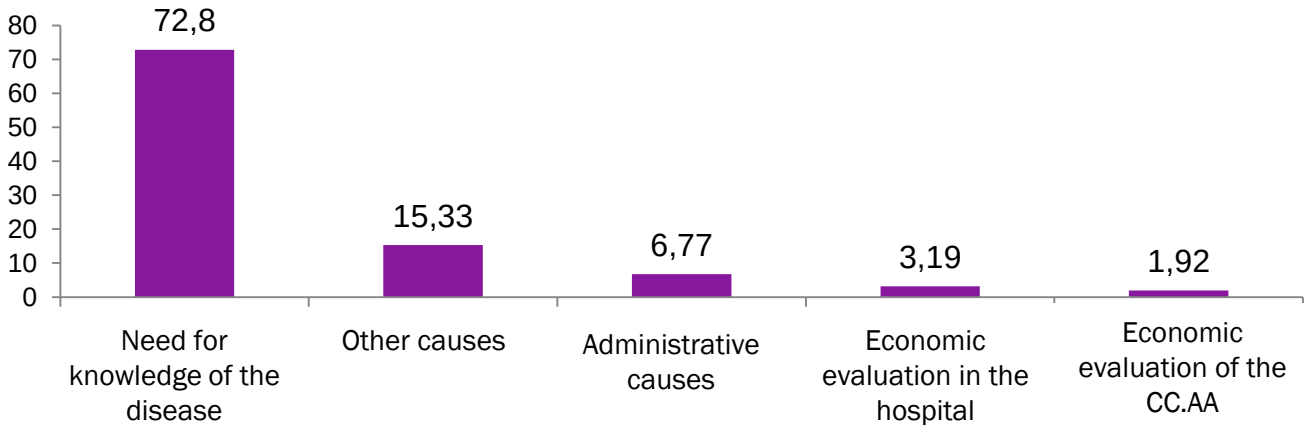

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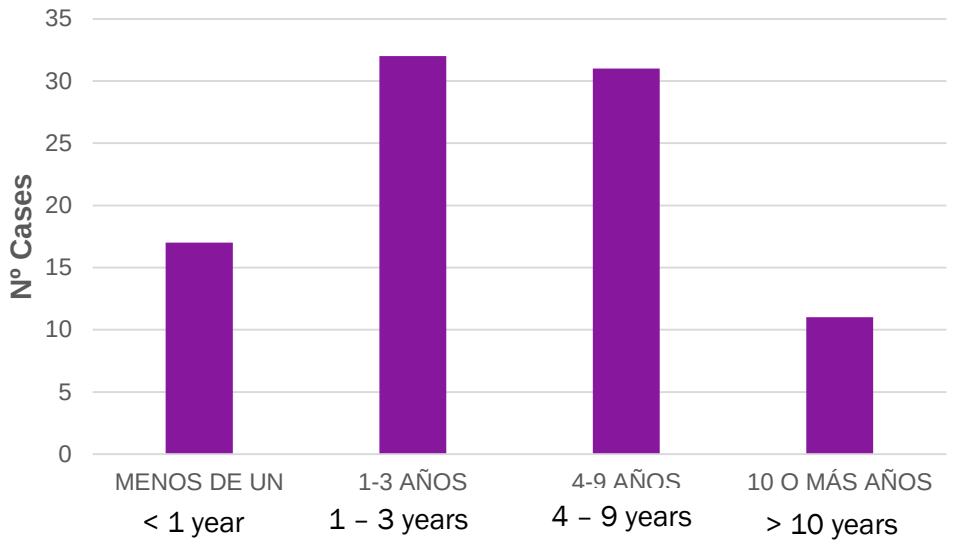
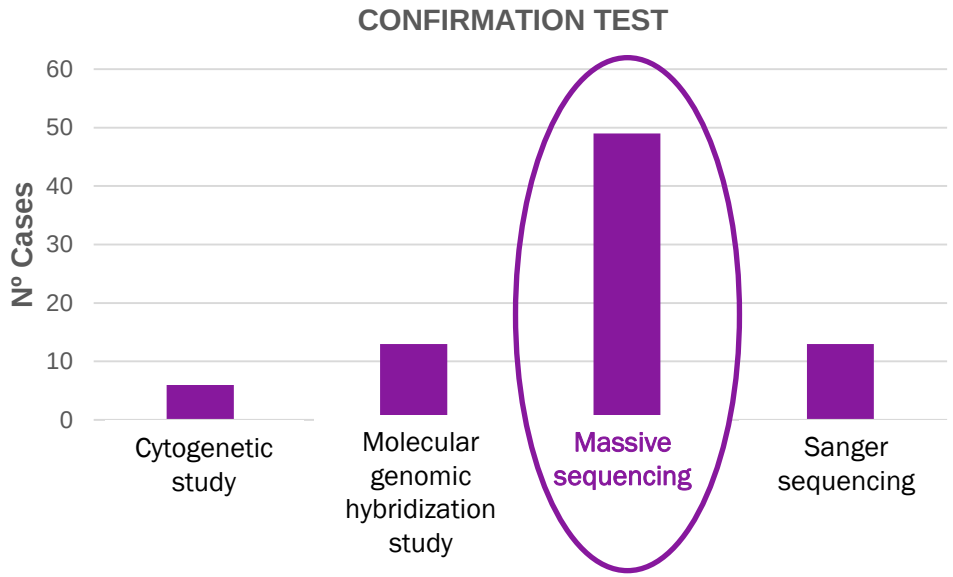
Diagnosis: Spain



Pilot Study: Comunidad de Madrid – Hospital Universitario de La Paz

State of the art

Diagnosis: Spain

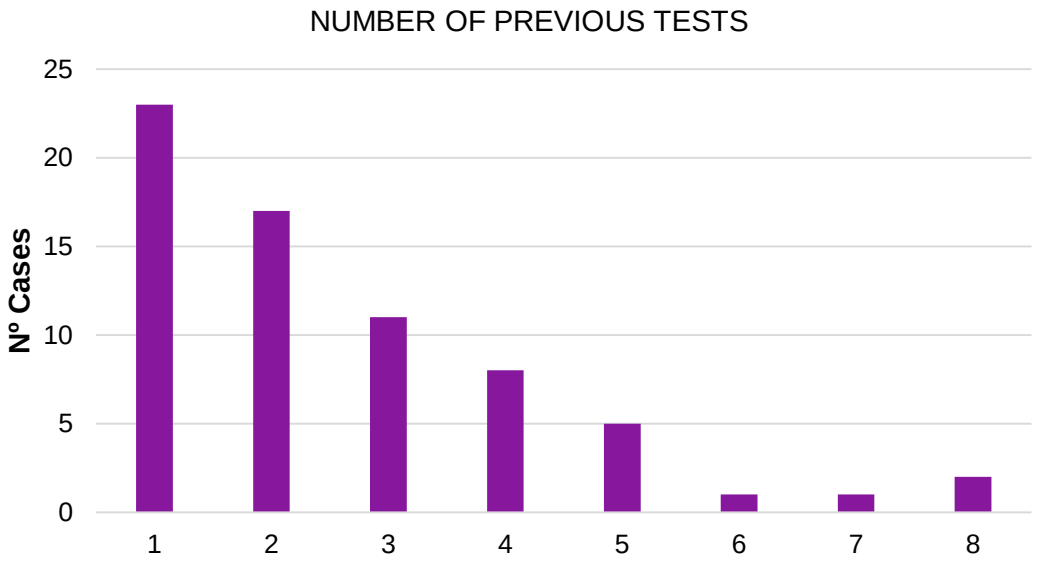


Pilot Study: Comunidad de Madrid – Hospital Universitario de La Paz

State of the art

Diagnosis: Spain

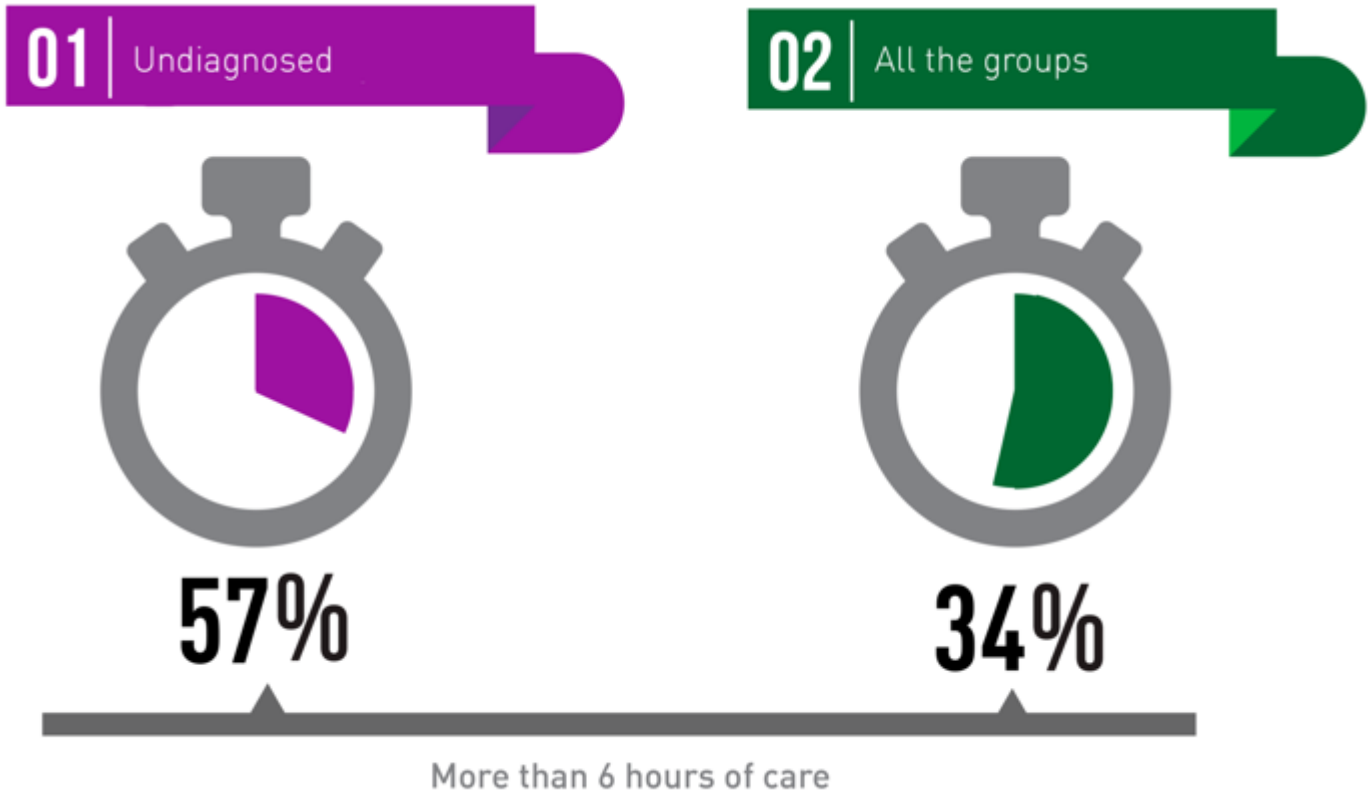
More than three quarters of the people have been made several tests before being referred to genetics to confirm the diagnosis.



Pilot Study: Comunidad de Madrid – Hospital Universitario de La Paz

State of the art

Diagnosis: Spain
Care



Analysis of the reality of the group of people without diagnosis (OBSER, 2019)

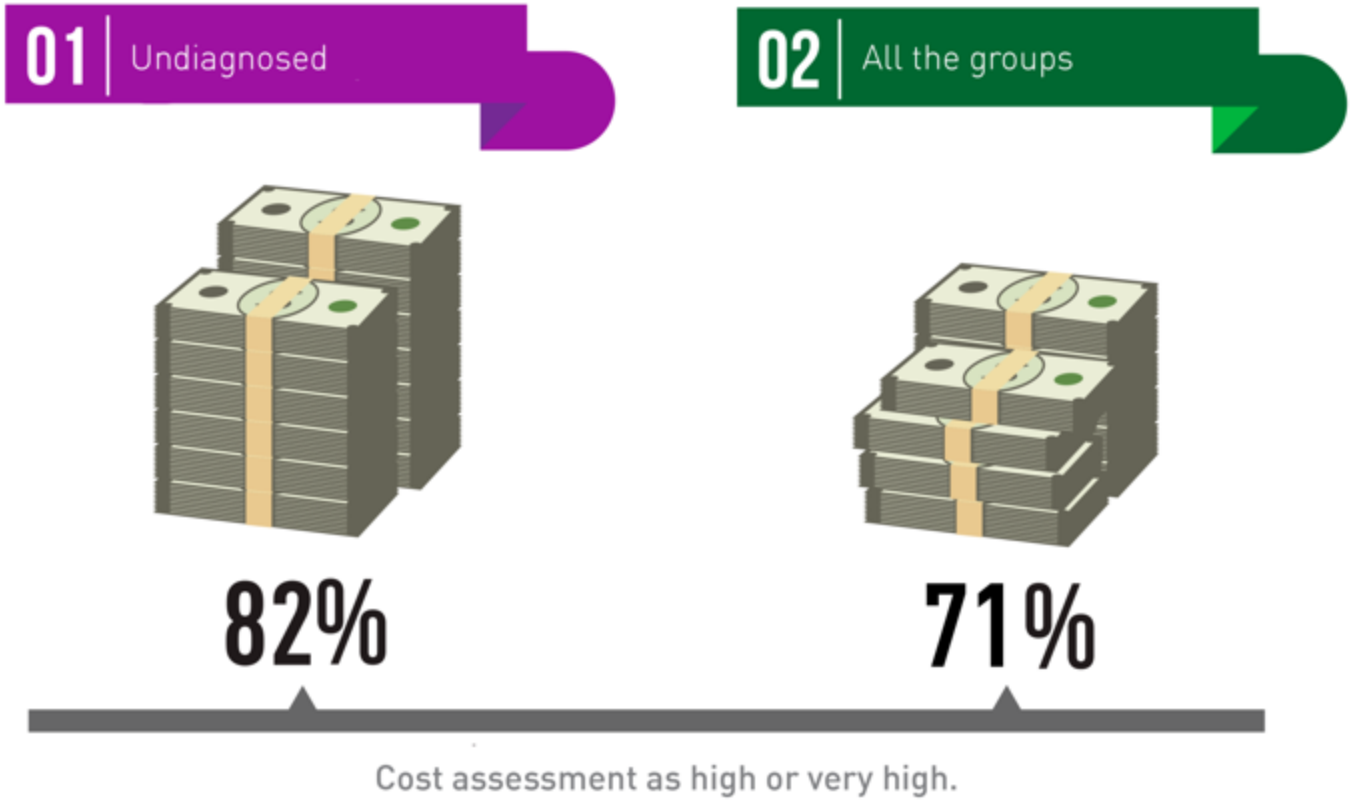
State of the art

Diagnosis: Spain
Care

Analysis of the reality of the group of people without diagnosis (OBSER, 2019)

State of the art

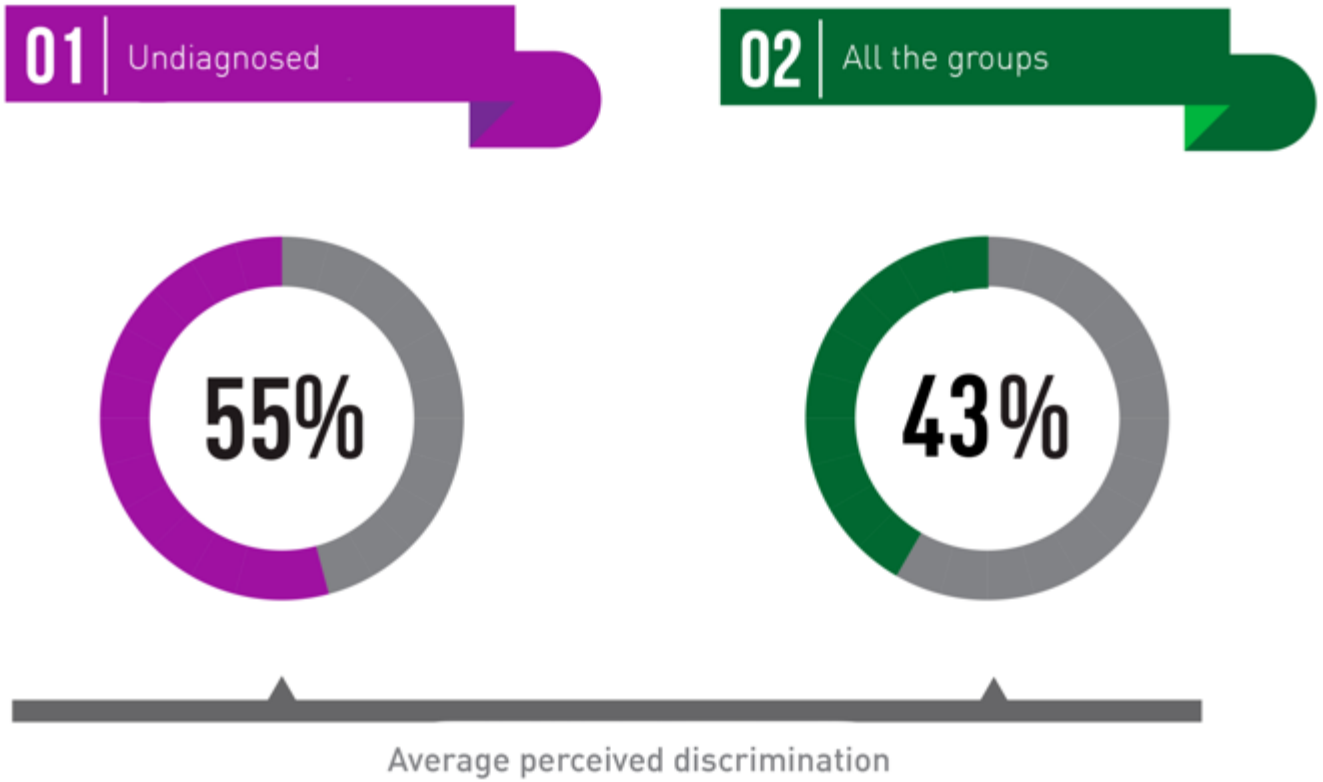
Diagnosis: Spain
Economic impact



Analysis of the reality of the group of people without diagnosis (OBSER, 2019)

State of the art

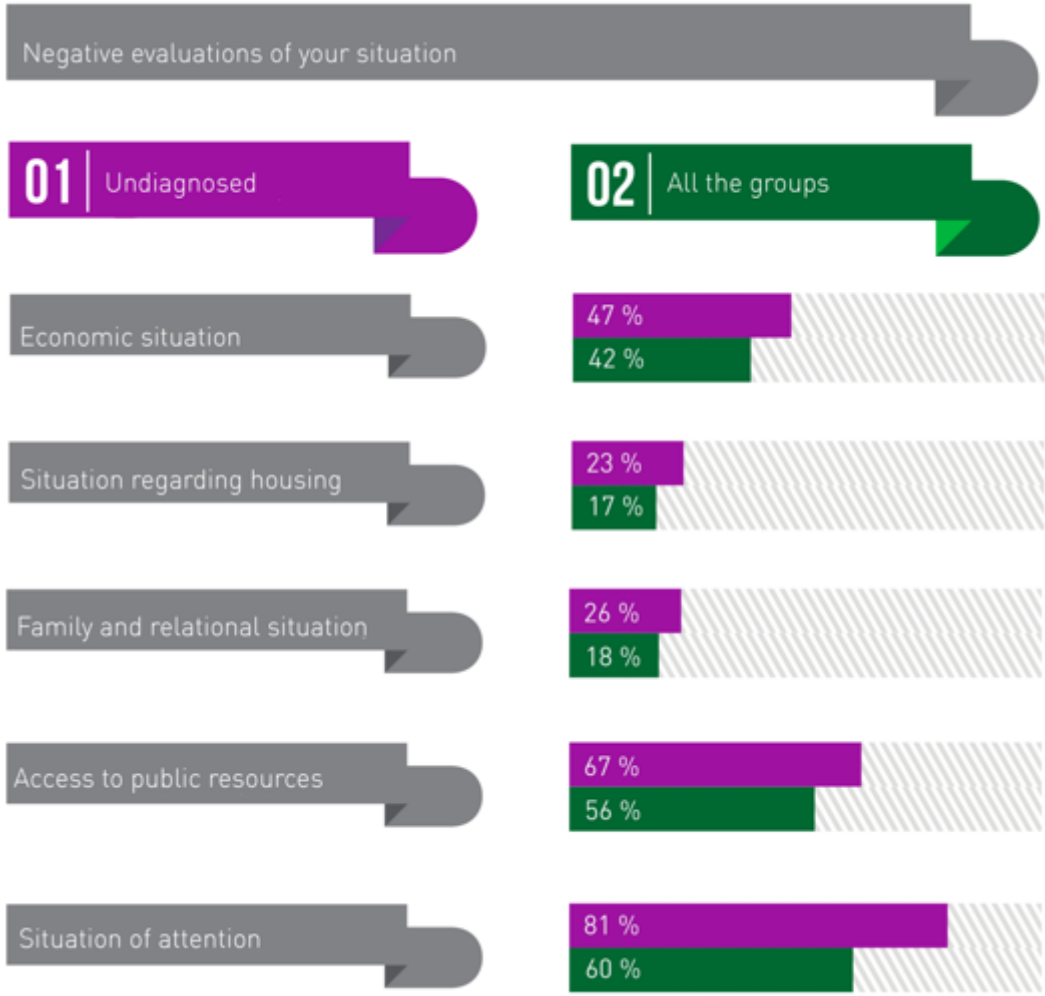
Diagnosis: Spain
Discrimination



Analysis of the reality of the group of people without diagnosis (OBSER, 2019)

State of the art

Diagnosis: Spain
Discrimination



Analysis of the reality of the group of people without diagnosis (OBSER, 2019)

State of the art

Diagnosis: Spain

- **Disinformation** about care and aid.

- **No financial aid**
  - Diseases "without name" not recognized.
  - Needs not covered in the basic portfolio (psychological care, rehabilitation).

- Costs to the system to cover the consequences.

Study on the situation of Social &Health Needs of Spanish People with Rare Diseases

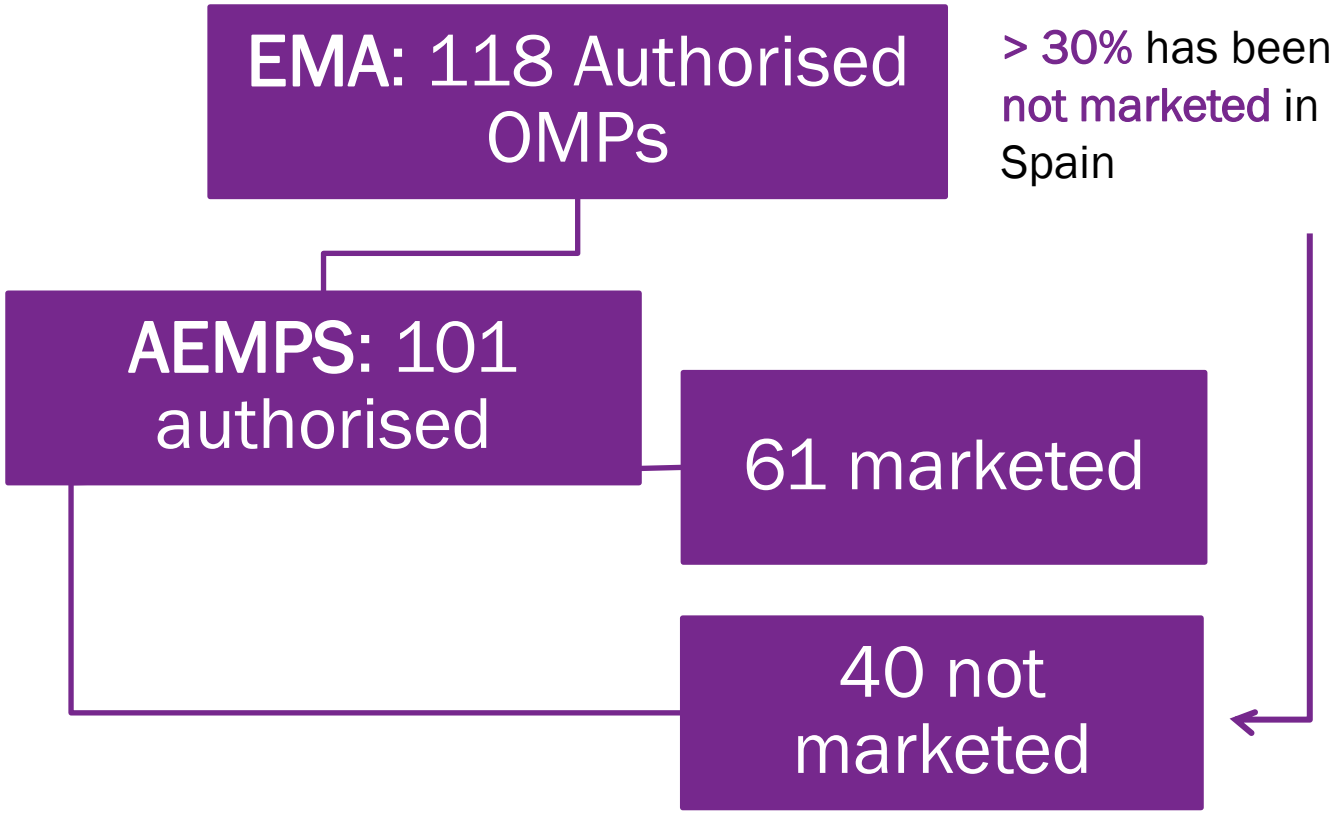
Misdiagnosis?

Effective treatment?

Genetic counseling?

State of the art

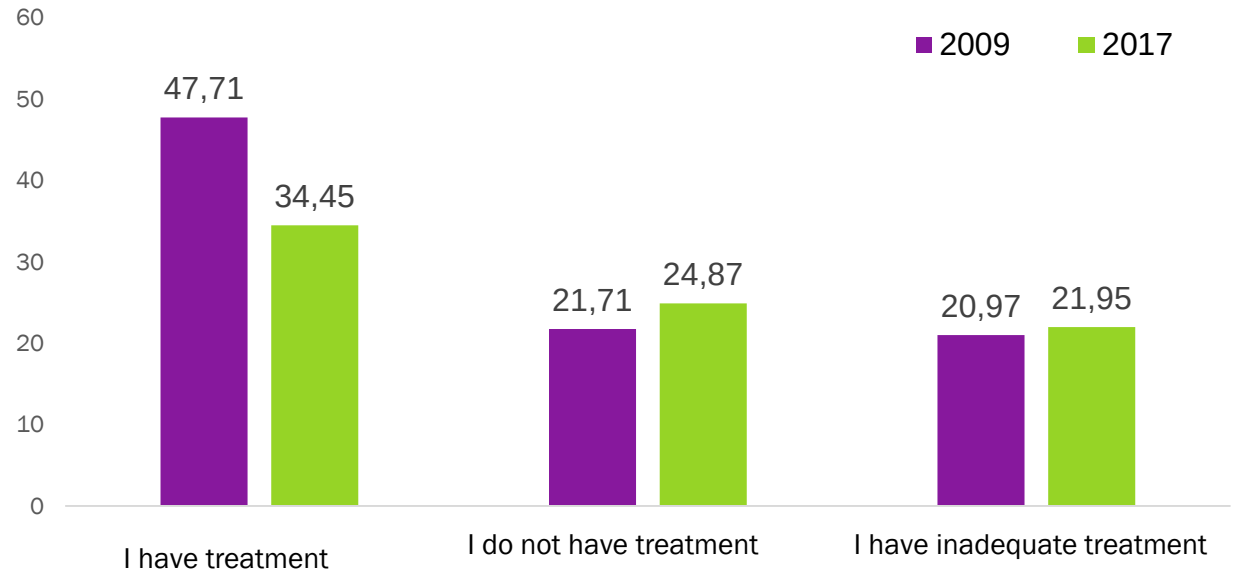
Treatment: Spain



State of the art

Treatment: Spain

- 34.45% of respondents assure that they received a customized treatment, **24.87% indicate they don't have it and 21.95% have a treatment, but consider it inaccurate**. In addition to these last two magnitudes, 46.82% of the sample is in a bad condition regarding their treatment.

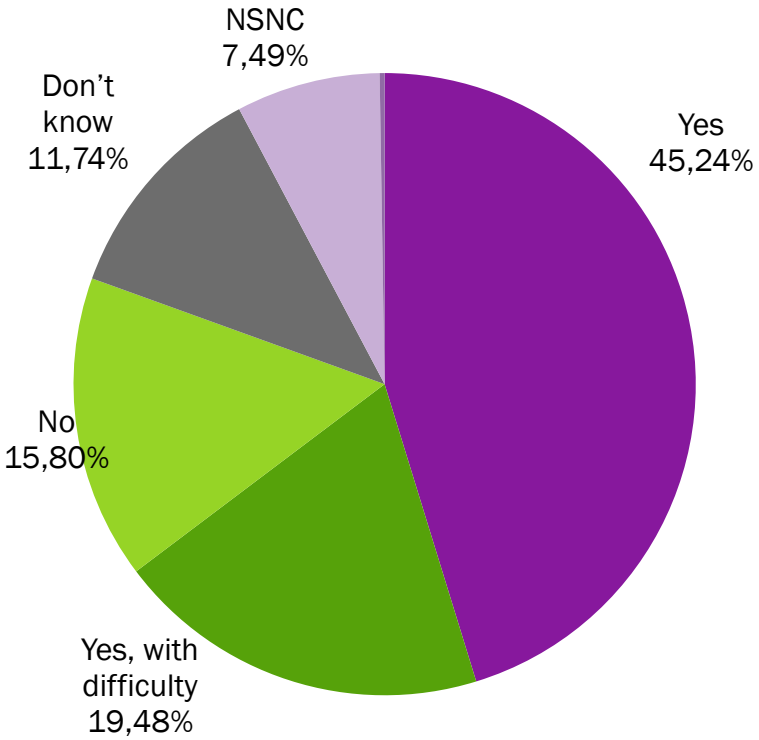


Study on the situation of Social &Health Needs of Spanish People with Rare Diseases

State of the art

Treatment: Spain

- **35.28% of respondents don't have access** to these products or purchasing them is utterly difficult.
- **45.24% of respondents have no monetary difficulty** to purchase these products.

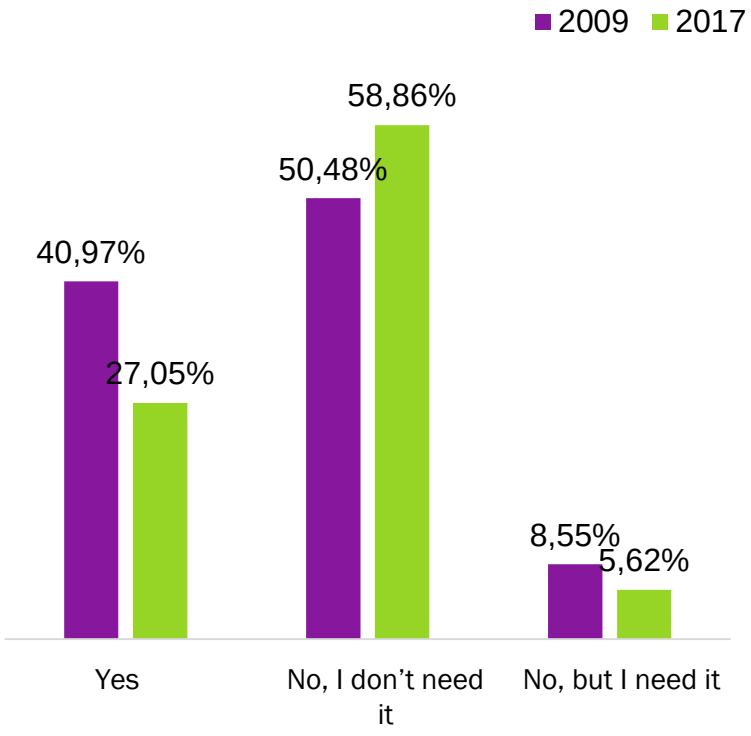


Study on the situation of Social &Health Needs of Spanish People with Rare Diseases

State of the art

Treatment: Spain

- **27% of people have been forced to move** to other Autonomous Communities to access a diagnosis, treatment or medication.



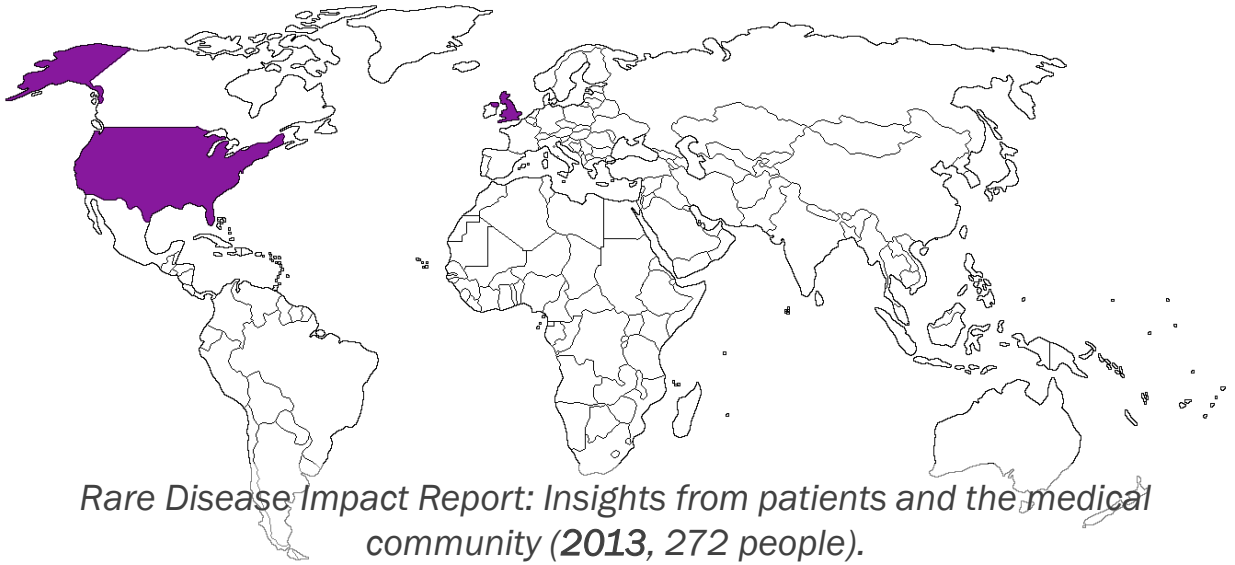
Study on the situation of Social &Health Needs of Spanish People with Rare Diseases

To global challenge
Global Solution

State of the art

USA, 2013

- According to patients surveyed, it takes **on average 7.6 years in the US** for a patient with a rare disease **to receive a proper diagnosis**.
- In order to get a proper diagnosis, a patient typically **visits up to 8 physicians**: 4 primary care and 4 specialists **an receives 2 to 3 misdiagnoses**.

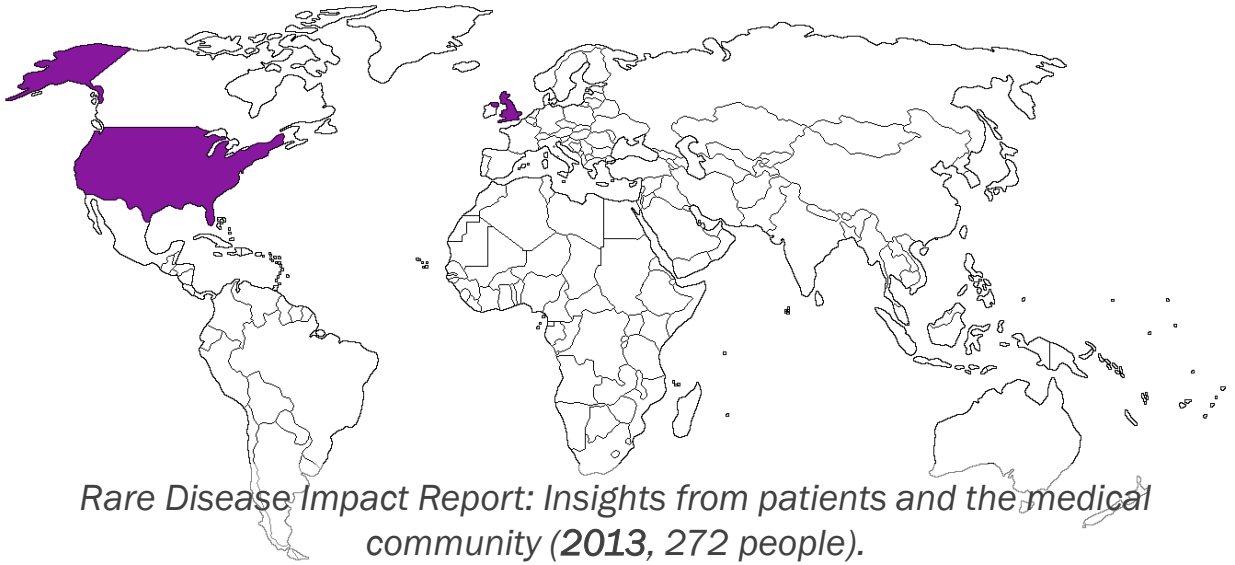


*Rare Disease Impact Report: Insights from patients and the medical community (**2013**, 272 people).*

State of the art

USA, 2013

- A majority of patients surveyed (60%) said there was an existing treatment for their rare disease.
- More than half of caregivers surveyed reported their loved one suffered from a rare disease for which there was no treatment (56%).



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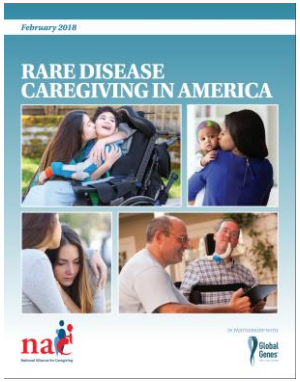
State of the art

USA, 2018

RARE DISEASES CAREGIVING IN AMERICA

Only 43% report there is a treatment available for the underlying disease or disorder

Most (82%) report that treatments or therapies are used for symptom management rather than actual disease treatment.



*Rare Diseases Caregiving in America (**2018**, 1406 care givers)*

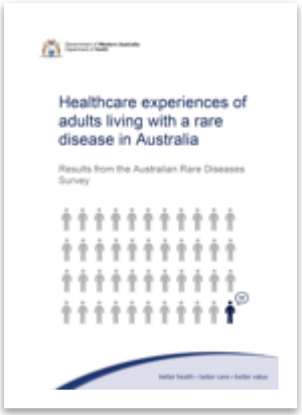
State of the art

Australia, 2014

- Nearly a third of respondents had to wait **5 years or more** to get a diagnosis.

Specifically:

- Less than 1 year: 49%
- 1-5 years: 21%
- 5-10 years: 21%
- 10-20 years: 9%

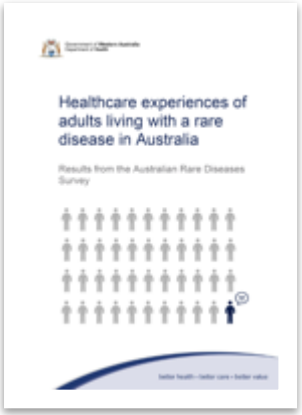


*Healthcare experiences of adults living with a rare disease in Australia (**2014**, 746 people)*

State of the art

Australia, 2014

- **45% of people got a wrong diagnosis.**
- More than half of respondents claimed to receive **information** on diagnosis, but **not enough.**
- **89% of people expressed their desire to participate in patient records** versus 20% who actually did.



*Healthcare experiences of adults living with a rare disease in Australia (**2014**, 746 people)*

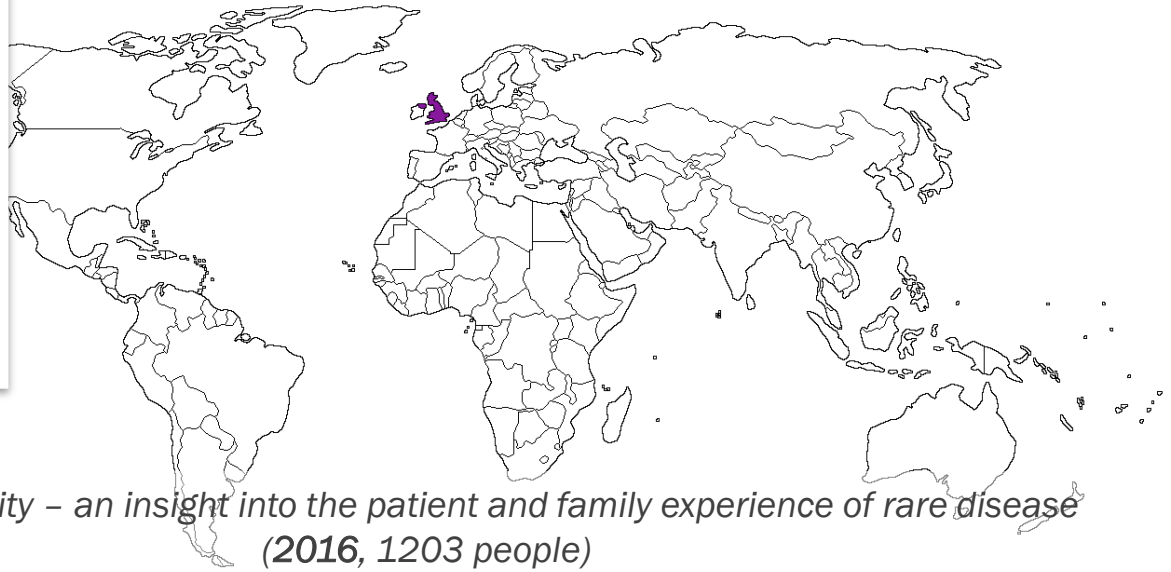
State of the art

UK, 2016

- The average rate of delay on diagnosis among respondents is **4 years.**

In particular:

- More than a year: 45%
- Over 5 years 25%, a quarter

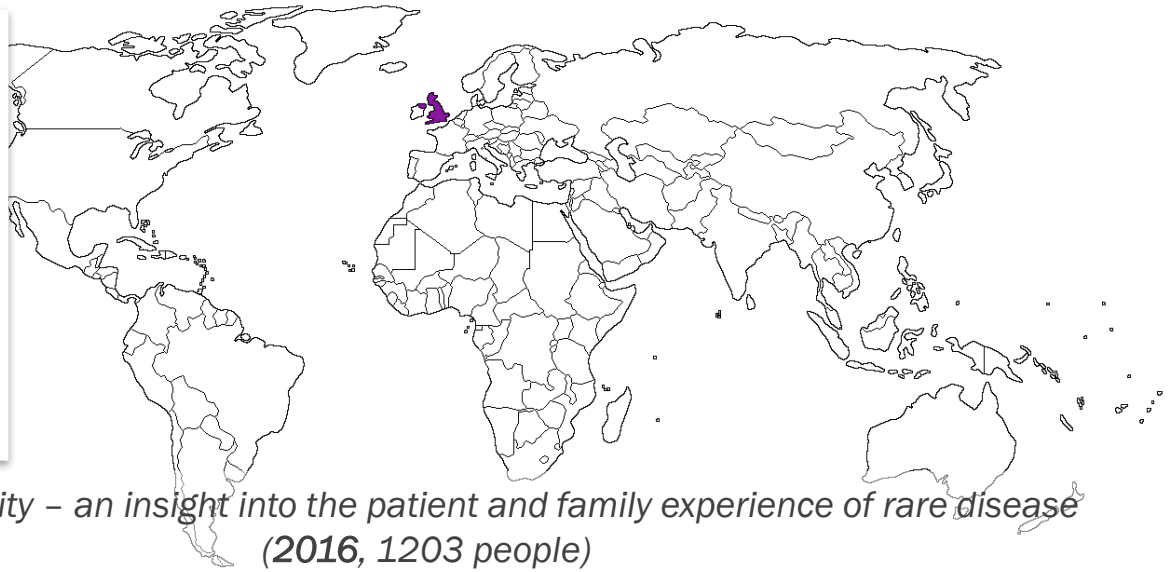


*The Rare Reality – an insight into the patient and family experience of rare disease (**2016**, 1203 people)*

State of the art

UK, 2016

- **45% declared having an authorized or approved medication** for their pathology, of which 87% declared being able to purchase it.
  - Thus 39% declared to have access to a medicine authorized or approved for their pathology.
- **From 20% of respondents who were informed about these drugs**, 55% had access to them.
  - 11% had access to this type of medication.



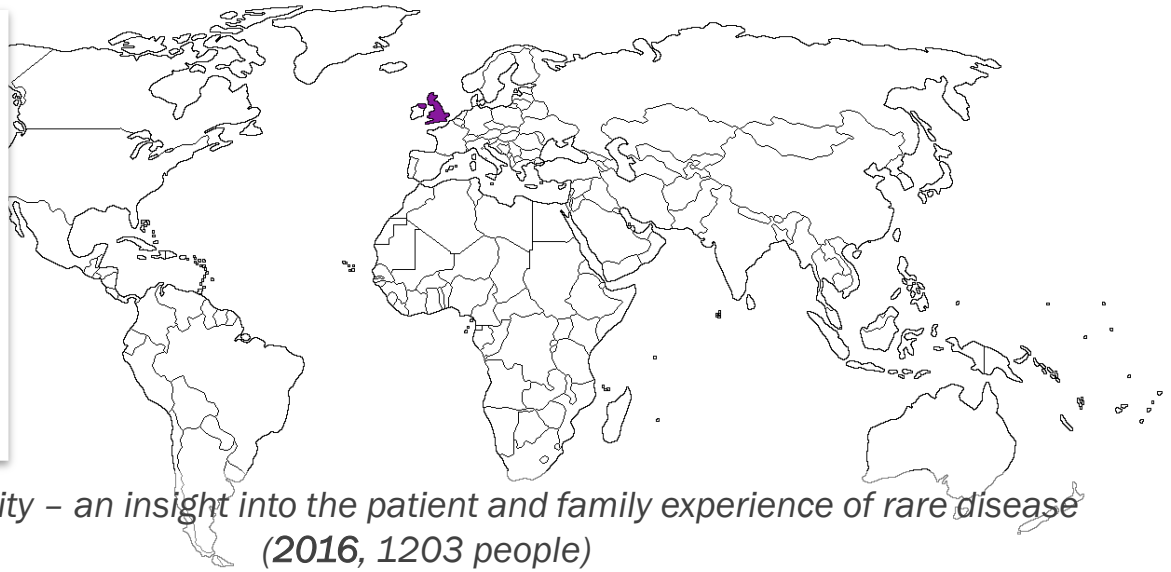
*The Rare Reality – an insight into the patient and family experience of rare disease (**2016**, 1203 people)*

State of the art

UK, 2016

TECHNICIANS:

- Average of 5 doctors to get a diagnosis.
- 71% saw 3 or more doctors and 10% more than 10.
- 52% claimed to receive an incorrect diagnosis before obtaining an accurate one.



*The Rare Reality – an insight into the patient and family experience of rare disease (**2016**, 1203 people)*

State of the art

WHO and UN involvement

- NGO Committee for Rare Diseases Launched a call for:

  - The integration of rare diseases into the upcoming landmark UN political declaration on universal health coverage (UHC), and

  - A UN resolution on rare diseases.

State of the art

WHO and UN involvement

Undiagnosed Diseases Network INTERNATIONAL

SWAN UK
syndromes without a name

SolveRD
Solving the Unsolved Rare Diseases

EURORDIS
RARE DISEASES EUROPE

RARE DISEASES INTERNATIONAL

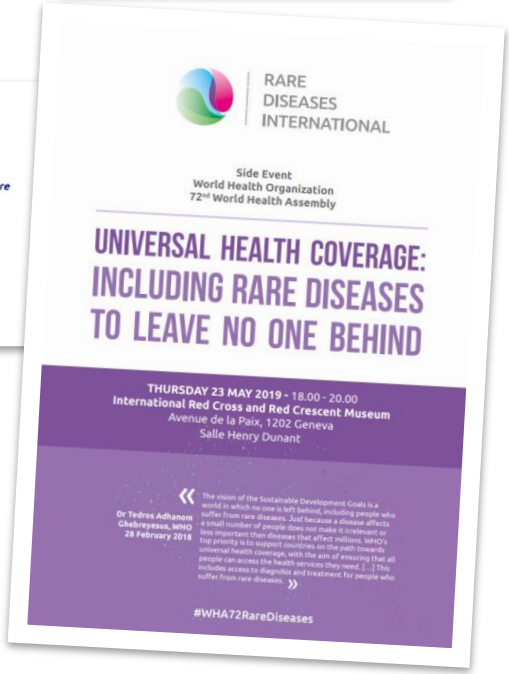
How Transformational Digital Technologies Can Contribute to Leave No One Behind in UHC: The Case of Rare Diseases

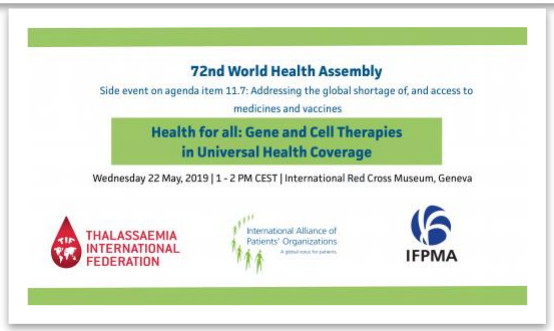
May 23 @ 12:30 pm - 2:00 pm

Organized by Permanent Missions of the European Union, Romania and Kuwait in collaboration with the Thalassaemia International Federation, Rare Diseases International (RDI), and the European Organisation for Rare Diseases (EURORDIS)

How Transformational Digital Technologies Can Contribute to Leave No One Behind in UHC: The Case of Rare Diseases

Thursday, May 23, 2019
12:30 PM – 2:00 PM





International Joint recommedations

The 5 Recommendations to address specific needs of undiagnosed rare disease patients

1. **Undiagnosed Rare Disease Patients should be recognised as a distinct population** with specific unmet needs by national authorities to enable development of personalised health and social care. Although some undiagnosed diseases are common, the vast majority are rare. Hence, in this paper we refer to undiagnosed patients as "undiagnosed rare disease patients".

2. **National sustainable programmes** dedicated specifically for undiagnosed diseases should be developed and supported by appropriate authorities in each country to enable rapid and equitable access to diagnosis and social support.

3. **Knowledge and Information sharing should be structured and coordinated** at national and international levels to optimise use of existing resources and facilitate access for all undiagnosed rare disease patients.

4. **Patients should be equally involved** with other stakeholders in the governance of undiagnosed diseases programmes and international networks to adequately address the priorities of undiagnosed rare disease patients and contribute to improved healthcare.

5. **Ethical and responsible international data sharing should be promoted** through existing initiatives to support diagnosis, increase clinical collaboration, facilitate research, and accelerate treatment of undiagnosed and rare conditions.

Why a Genomic Strategy?

Network and coordinated action

Why Genomic Strategy?

Pearl Borau. The objective was the implementation of a Strategic Plan on genomic medicine in order to achieve greater **equity** and quality in the public health system.

El Senado da luz al Plan de Genómica...

● La Comisión de Sanidad propone dotar la estrategia en genómica con financiación finalista

● Para el desarrollo de plataformas en red apuestan por los recursos del Fondo de Cohesión

Tras un año de sesiones semanales en el Senado, la Comisión de Sanidad de la Cámara Alta ha finalizado la Ponencia de Estudio sobre Genómica, y lo ha hecho, alcanzando un acuerdo unánime.

Las formaciones políticas han consensuado un documento con trece recomendaciones que será debatido próximamente en el Pleno del Senado y que pretende ser la 'escaleta' de la futura Estrategia Nacional de Medicina de Precisión en la que también trabaja ya el Sanidad y las comunidades autónomas.

Los senadores proponen el desarrollo de esta estrategia a diez años. Si bien tendrá que elaborarse anualmente un informe de evaluación de resultados que deberá presentarse al Consejo Interterritorial.

La financiación, como han ido pidiendo en el último año los diferentes expertos que han conformado la ponencia, deberá ser finalista. Para el trabajo en red, el Senado propone aprovechar los recursos existentes y financiarlo, entre otras fuentes, a través del Fondo de Cohesión. P5

... Pero Hacienda y Sanidad no lo tienen en cuenta en los PGE

El Senado apunta a la financiación por parte del Fondo de Cohesión pero esta vía recibe la misma dotación que el año pasado, unos escasos 3,7 millones de euros. P6-7

La ministra de Sanidad, María Luisa Carcedo, explicó la semana pasada los presupuestos que tendrá que gestionar su cartera si finalmente se aprueban las cuentas.

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Gaceta Médica

Why a Genomic Strategy?

Background: 2015

Feder quiere un plan específico para pacientes sin diagnóstico

MADRID
REDACCIÓN

El presidente de la Federación Española de Enfermedades Raras (Feder), Juan Carrión, pidió ayer un programa específico para pacientes sin diagnóstico que establezca "itinerarios asistenciales rigurosos" que comprendan análisis clínicos, segunda opinión médica, análisis de laboratorio, cho mayor".

Un retraso del diagnóstico supone que, en el 30 por ciento de las ocasiones, la enfermedad se agrave, añaden los responsables. Las principales causas de este retraso, dicen, son la escasez de pruebas de detección precoz financiadas por el SNS, así como el desconocimiento de la especialidad en el acceso a los tratamientos.

En concreto, solicitan que el programa específico para los pacientes establezca los criterios para la recepción de los casos de dificultad diagnóstica; implantar las medidas necesarias a nivel de los departamentos de genética, cribado neonatal, atención primaria, especializada y centros, servi-

24 Política

23 de febrero al 1 de marzo de 2015 GM

Feder solicita un programa específico en las CC.AA. para personas sin diagnóstico

Esta situación hace que un 30% de los pacientes vean cómo se agravan los síntomas de su enfermedad

C.S.
Madrid

La Federación Española de Enfermedades Raras (Feder) ha registrado más de 3.160 consultas sobre casos sin diagnosticar en España. Con esta cifra como punto de partida, la directora de la Federación, Ana Anconchea, explicó que se estima un promedio de cinco años entre que aparecen los primeros síntomas y el diagnóstico, pero un 20 por ciento tarda hasta 10 años en recibirlo.

Según el estudio sobre la situación de las Necesidades Sociosanitarias de las personas con EE.RR. en España (UNNERo), más del 40 por ciento de las personas que tienen un retraso diagnóstico no reciben apoyo ni tratamiento. El 27 por ciento lo reciben equivocado y un 30 por ciento tienen consecuencias en el agravamiento de su enfermedad.

El verdadero problema no es solamente la falta de diagnóstico por ausencia de registros o de investigación en enfermedades de baja prevalencia, sino que cuando se dejan de diagnosti-

Para poner de relevancia la falta de equidad que existe entre CC.AA.; Feder, en coordinación con la Organización Europea de EE.RR. (Eurordis) y con la Alianza Iberoamericana de EE.RR. (Aliber), ha organizado una serie de actividades que tendrán lugar entre febrero y marzo a propósito del Día Mundial.

Gestos por las EE.RR.

El objetivo de esta campaña es, según Carrión, sensibilizar y tener presentes las necesidades de las personas que padecen estas patologías. Para ello, se realizarán más de 300 actos en toda España. "Actividades que cuentan con el apoyo de más de 283 organizaciones y de empresas que han querido colaborar para que estas necesidades se escuchen a una única voz".

Los actos principales serán: la III Jornada de Expertos que se celebrará el 20 de febrero en el ministerio de Sanidad para posibilitar que profesionales y representantes de asociaciones de enfermedades poco frecuentes se den cita y analicen las necesidades de este colectivo y sus

Why a Genomic Strategy?

Network and coordinated action

2015: MSSSI announces the development of the PILOT PLAN FOR DIAGNOSIS after our campaign on diagnosis.

2017: CISNS approved 800,000 € TO IMPLEMENT THE PILOT PLAN.

2018: there are CCAA not involved.

» 11 Autonomous Communities present the documentation for the execution of the plan.

2019: We will work together with Hospital Universitario La Paz and CIBERER identifying care routes that favor access to diagnosis.

Sanidad añadirá 800.000 euros para investigar patologías raras

AGENCIAS ■ Madrid

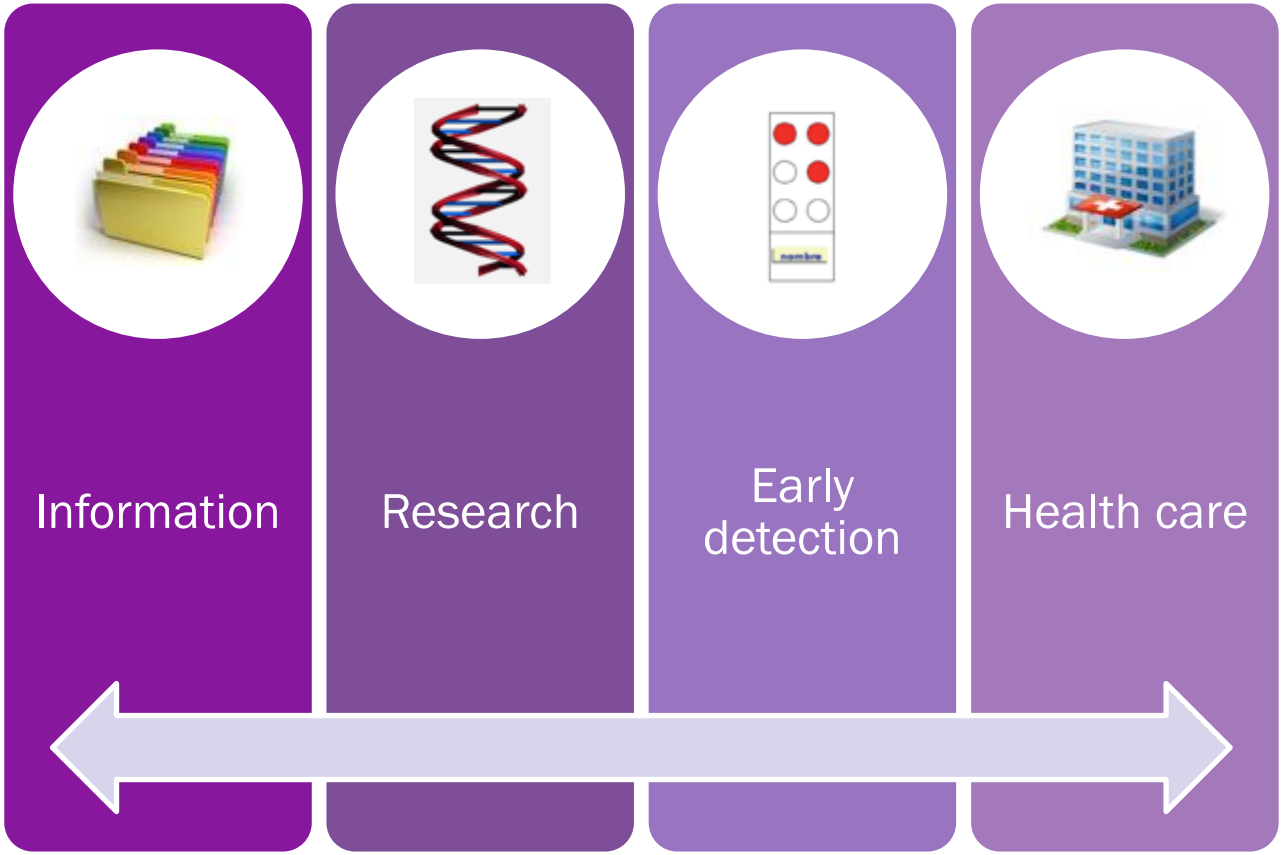
El Ministerio de Sanidad hará una aportación adicional de 800.000 euros, que se suma al millón ya distribuido entre las comunidades autónomas para la investigación de enfermedades raras, para poner en marcha una prueba piloto que pretende agilizar el diagnóstico.

Con esta partida se preten-

feder
entidad de utilidad pública
FEDERACIÓN ESPAÑOLA DE ENFERMEDADES RARAS

Beyond Genomics

Whole Strategy



Whole Strategy

Beyond Genomics

The elaboration of a Strategy in Genomic, Personalized and Precision Medicine for the SNS **is considered necessary**.

- And the participation of patients.

The Strategy should take advantage of and optimize existing resources and initiatives in development by the Autonomous Communities and the State Administration, as well as establish a network that should implement a plan for its optimization and expansion, as well as be financed, among others, **with resources of the Health Cohesion Fund**.

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IV. OTRAS ACTIVIDADES PARLAMENTARIAS

PONENCIAS DE ESTUDIO CONSTITUIDAS EN EL SENO DE LAS COMISIONES

Ponencia de estudio sobre genómica, constituida en el seno de la Comisión de Sanidad, Consumo y Bienestar Social (antes denominada Comisión de Sanidad y Servicios Sociales). (543/000006)

INFORME DE LA PONENCIA

A la Excma. Sra. Presidenta de la Comisión de Sanidad, Consumo y Bienestar Social.

Excma. Sra.:

La Ponencia de estudio sobre genómica ha aprobado, por unanimidad, en su reunión celebrada el día 15 de enero de 2019, el Informe que se acompaña como Anexo.

Palacio del Senado, 15 de enero de 2019.—Nerea Ahedo Ceza, Antonio Alarcó Hernández, María del Carmen de Aragón Amunárriz, Anna Azamar Capdevila, María Perla Borao Aguirre, Yaiza Castilla Herrera, José Martínez Olmos, María Concepción Palencia García, Maria Teresa Rivero Segalàs y Alfonso Jesús Rodríguez Hevia González.

ANEXO

INFORME DE LA PONENCIA DE ESTUDIO SOBRE GENÓMICA

I

INTRODUCCIÓN

1. Antecedentes.

La creación en el Senado de la Ponencia de estudio sobre Genómica, en el seno de la Comisión de Sanidad y Servicios Sociales, hoy denominada de Sanidad, Consumo y Bienestar Social[1], fue el resultado de una moción consecuencia de interpelación presentada por el Grupo Parlamentario Socialista, debatida en la sesión plenaria celebrada el día 8 de marzo de 2017 y aprobada en dicha sesión por unanimidad, con la incorporación de una enmienda del Grupo Parlamentario Popular en el Senado.

1 Dicho cambio de denominación se llevó a cabo por reforma del Reglamento del Senado, aprobada por el Pleno de la Cámara en su sesión de 19 de junio de 2018 (BOCG, Senado, núm. 256, de 28 de junio de 2018).

cve: BOCG_D_12_341_2574

- Ensure that this optimization can be implemented in a sustainable manner.

Beyond Genomics

Whole Strategy

The Strategy should incorporate objectives to ensure the genomic sequencing of **patients with rare diseases** and their relatives.

- *Recognition.*

The **creation of the specialty of Clinical Genetics** and the incorporation of professionals is recommended.

- *Historical recommendation.*

The Strategy should have a **Coordination and Management Committee** and will create a Monitoring Observatory.

- Patient Committee?

Patient Engagement

ENSERio Study

Patient Engagement

- Second phase of the ENSERio Study: analysis by CCAA of diagnosis and treatment.

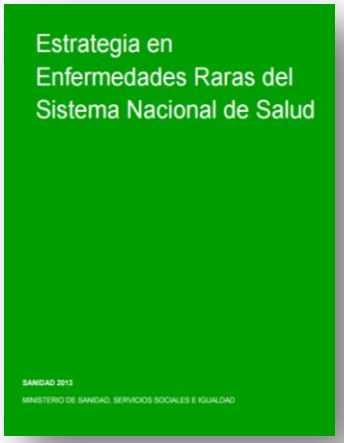
At this time we have come to cover the share of sample representation in 11 Autonomous Communities (6 already represented and 5 achieved in 2019) and will begin to analyze the results in the coming months.

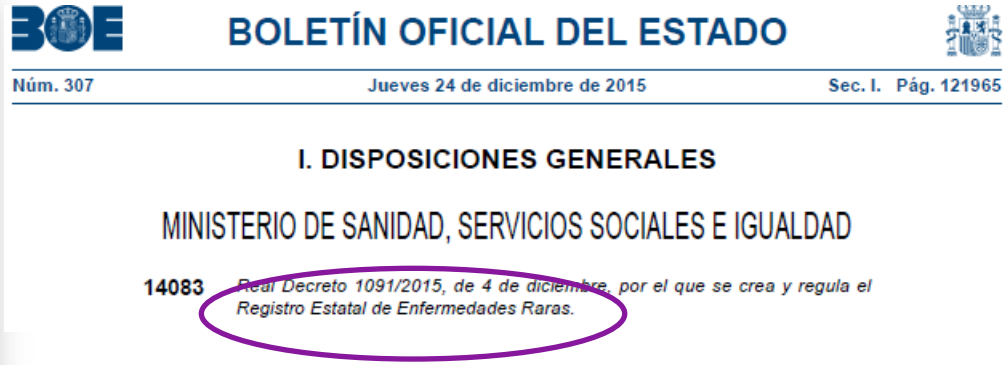
- Study of diagnostic delay through clinical data

- CETF Group for community participation in the European SOLVE RD project

Committees in which we are represented

Patient Engagement

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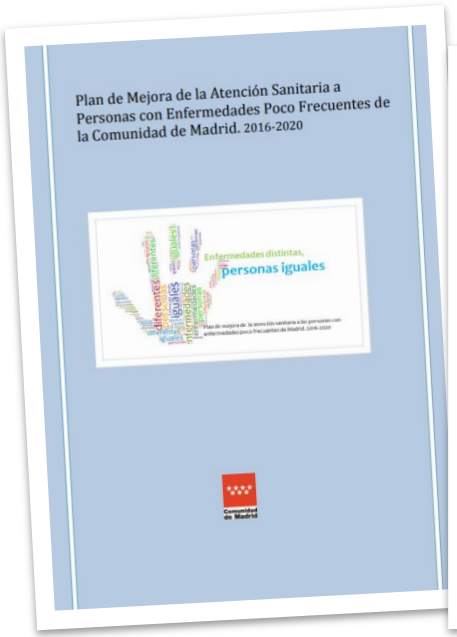
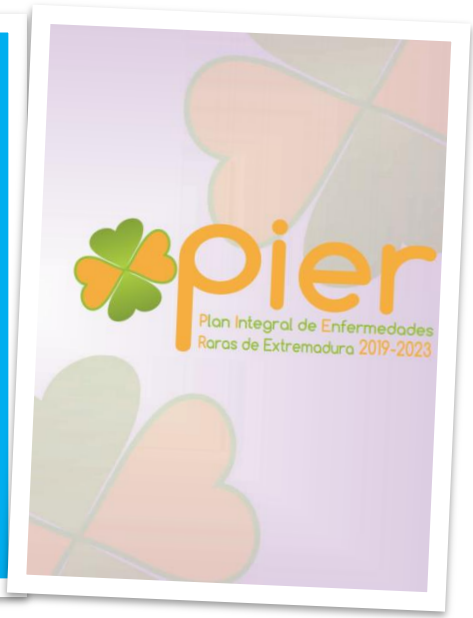
I. DISPOSICIONES GENERALES

MINISTERIO DE SANIDAD, SERVICIOS SOCIALES E IGUALDAD

14083 *Real Decreto 1091/2015, de 4 de diciembre, por el que se crea y regula el Registro Estatal de Enfermedades Raras.*

FEDER participates in the working group on **CSUR**





Our proposals

Our Prioriities

1. Promote a Rare Diseases National Strategy Implementation Plan
2. Assure sustainability of Rare Diseases National Register
3. Promote research on Rare Diseases
4. Promote education and information on Rare Diseases
5. Strengthen Expertise Centers designation
6. Introduce a RD global assistance model
7. Achieve an equal and fast access to diagnosis and treatment
8. Promote laboral inclusion for Rare Diseases
9. Encourage education inclusion for Rare Diseases
10. Strengthen social services specially those related to Disability and Dependence

Our proposals: 2019

❖ Promote that the National Plan of Genomics and Precision Medicine have express reference to the RD:

- Request the Ministry of Health that the development of the State Plan of Genomics and Precision Medicine to which it has committed has express reference to the RD.

- SoA of the Plan and its implementation.

- Propose initiatives to the Parliamentary Groups.

- Request the Ministry the participation of patients in the corresponding forums for designing the Plan.


Acto

FEDER pide la creación la especialidad de genética clínica en España

La Federación Española de Enfermedades Raras pide más investigación y que se autoricen más medicamentos

La Federación Española de Enfermedades Raras (FEDER), Juan Carrión quiere que se implante la especialidad de genética clínica en los hospitales españoles. Así lo reivindicó ayer el presiente de la FEDER, Juan Carrión, en un acto celebrado en el Palacio de San Esteban con motivo del Día Mundial de las Enfermedades Raras, que se conmemora hoy. En este marco, Carrión recordó que España es el único país de Europa que no cuenta con esa especialidad «crucial en la investigación sobre este tipo de enfermedades».

Más fotos

Patient Engagement


Extraordinary problems
Extraordinary solutions