Síndrome de
RETT
ASOCIACIÓN ESPAÑOLA
www.rett.es

ACTIVITIES REPORT
2015

INDEX

PRELIMINARY ISSUES

ORIGINS AND RATIONALE

AESR, Spanish Rett Syndrome Association, is a nonprofit association created by parents of children affected by this disease. It is a National Association, independent, non professional, with juridical personality, engaged with the self-help of the affected families and creating awareness in society.

The reason for our existence is to improve the quality of life of our girls, and the protection of the rights and equal opportunities for our daughters, as we believe that they are in a huge disadvantage compared to their peers of the same age.

RETT SYNDROME

Rett syndrome (RS) is a rare disease (RD). As RD is believed that could affect 1 in 10-12,000 girls .The RS is a serious genetic neurodevelopment disorder, that occurs almost exclusively in girls as it is originated in the X chromosome.

It was first described in 1966 by Dr. Andreas Rett in Vienna and is in the most recent times when we are beginning to understand the complex mechanism of appearance and involvement of this disease.

We now know that the origin of RS does not follow Mendelian classical laws of genetic transmission, it is rather an epigenetic disease in which a mutation or deletion in a gene does not alter the production of a single protein, but that mutation or deletion affects the configuration and expression of many other genes, and thus affects the production and appropriate expression of important proteins in human development.

According to DSM IV-R, Diagnosis of Mental Disorders System, which is being used today, the RS is a Pervasive Developmental Disorder, independent of ASD (autism spectrum disorders), and the only TGD that has a distinct and known biological basis. As explained below, in most cases, there is an altered gene, MECP2, which causes changes in adequate production of MeCP2 protein that is modulating / regulating other genes and have great importance in the neurodevelopment.

Today we can say, in the words of Dr. Marian Pineda that the RS is a "SINAPTOPATHY". Girls with Rett Syndrome cannot generate adequate synaptic connections, necessary for proper brain organization, due to the failure in the expression of MeCP2. It is known that girls with RS, at an anatomical and pathological level, have shorter neurons, weaker dendrites and far fewer connections between them. This is an indicator of poor maturation, deficits in synaptic connections, poor neuronal plasticity that significantly affects development and learning in general.

RS is not a degenerative or regressive disorder. Although one of the symptoms is the loss of acquired functions, professionals classify it as a Developmental Disorder. It is therefore a complex developmental disorder that goes through different stages or phases. Despite the absence of data on life expectancy, it is believed that it is normal to exceed 40 years.

The RS is a rare disease, but one of the most numerous and known today and the leading cause of intellectual disability in women today. It affects women of different socio-economic nature, and every racial and ethnic group and is present in everyone similarly. There are reports of males with RS, but for statistical reasons and lack of conclusive studies, we always talk about a disease closely linked to the female gender.

We believe that in Spain there may be over 2,000 diagnosed cases and many more undiagnosed adult women. In our association, we currently have about 300 girls / Adult diagnosed with RS. According to our information, there is no government statistics on the incidence and number of affected women in Spain.

So when we talk about Rett syndrome, we refer to a serious deterioration in child neurodevelopment, which affects girls in their whole being, causing a significant intellectual , motor, communication and sensory dissabilities:

- Motor Limitation: there is a wide range that goes from the total dependence on others when there is an absence of voluntary mobility to awkward or retarded movements of milder cases.

- Limitation in communication. Their ability to express themselves through language is also quite null, so it is necessary to use augmentative and replacement techniques to communicate with them.

- Sensory Limitation. They are frequent problems in the balance, vision, hearing and touch, which makes difficult their knowledge of the environment.

- Medical need. The vast majority have chronic pharmacological treatment for their condition. Many suffer epileptic seizures in different degrees and frequencies.

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all in all, we talk about girls with high dependency, who need continuous support of others to perform basic and instrumental activities of daily living due to a lack or loss of physical, mental, intellectual and sensory autonomy.

AESR SET UP

AESR MISSION

- Protection of rights and equal opportunities for our daughters.

- Appeal for a qualified medical assistance . In order to do this we must promote research on the syndrome at all levels, promote interdisciplinary collaboration, informing Society and promoting specific training.

- Appeal for a quality educational care, and also promote research and fight for the provision of materials, training and tools.

- Ensure the Socio-Economic benefits relying on the laws of the country.

AESR VISION:

- We aspire to have the essential needs overlayed at every stage of the disease without continuous struggle. And also to assure a continuity, both in quantity and quality of: Early-education, special attention and adult-care centers.

- We pretend to have an extensive knowledge about health, welfare and educational sector concerning Rett Syndrome..

- We want to be a reference in advisory issues at all stages of the disease and participate in the research committees.

- We long to turn into a large Association, both in number of partners and actions, with activities for both children and parents and care givers. With open and stable relations at work with all partners related to Rett Syndrome.

- Clear protocols and adapted healthcare

- Full accessibility.

OFFICES

The AESR is a national association with headquarters in Valencia. It is structured by delegations, and these are an important tool to accomplish the misión.

The delegations are formed by parents of girls with RS and distributed by zones, this division generally coincides with the province limits. They are responsible for contacting the local families, answer questions in difficult times; as well as inform the families of the available professionals and entities related to Rett syndrome. They also promote and seek both central and local resources for Rett girls.

Currently AESR, has the following offices:

Delegation of Madrid

Delegation of Valencia

Delegation of Badajoz

Delegation of Toledo

Delegation of Valladolid

Delegation of Pontevedra

Delegation of Málaga

Delegation of Granada

Delegation of Seville

LINES OF ACTION

targeted to families

Reception

This activity consists in offering emotional support to new families, especially to the main caregiver. It is carried out first by the social worker who will enable the contact with other families in the same area where they live. It is usually the delegate who will offer the first support among the peer group.

This support will be made first by phone and then, if the family is interested, through meetings over time with the group of families in their area.

Once a year all the Rett families in Spain, are called to the Family Meeting where new families are welcomed, propitiating spontaneous contact groups and self-help.

Results: During this year we have welcome 25 new Rett Families

Information and Guidance

This activity is to provide information about the entity as well as Rett Syndrome, families, professionals and volunteers. It is a first contact with the Association. Usually the first contact is made by mail or phone.

Families, employees and professionals are usually informed by mail of every new issues and events.

The results of this activity has been as follows:

33 new partners.

13 professionals (pedagoges, physiotherapists, psychologists and Centers)

30 students (Bachelor, Teachers, Physiotherapists, Psychology, Language therapy Occupational Therapy, Primary Education, Social Education and Social Work) all performed academic research on RS.

10 media, mainly radio and press, both print and online

Specific and individualized intervention. Social Worker.

The family is contacted by the Association and the social worker will be the one that makes the diagnosis of the needs presented by the family and report and provide appropiate guidance for each case, making a follow up.

This activity is performed by the professional by telephone, email or personal interview.

The results were: 45 families were advised on their demands on topics related to Disability Dependency,Tax benefits and economic benefits for the care of children with chronic disease.

Rett Annual Meeting of Families

This is an Annual Meeting where all familiies with children with Rett Syndrome can assist. Professionals in different areas will inform members and families, with lectures related to care, rehabilitation, legal and administrative matters. Research news are also reported annually in this meeting

This year we celebrated the "IX ANNUAL MEETING OF FAMILIES RETT 2015" May 30-31 in Madrid at Hotel Novotel Puente de la Paz. Result: Attendance of 45 families

Asistentes al IX Encuentro de Familias 2015

Girls performing Educational ludic activities. Organized by the Circus Piruleto volunteers this year our children s also enjoyed music therapy

Family gatherings in the delegations

(León) Marzo

These are meetings of families of the same area, The idea is to spend the day together to share experiences among the peer group. They say their goal is "to get an injection of optimism in this hard road we take with our girls but that so often give us a meaning for life. "

TARGETED TO GIRLS

Happy Birthday!!!!

The association congratulates all girls on the day of their birthday, through a personalized card and a small gift.

IRENE

MARIA

Music Therapy

This activity, has been fulfilled through this year, it aims to improve the physical, social, communicative, emotional and intelectual aspect in RS through music therapy. We know that our girls show great receptivity to music, that is why music becomes an essential element to reach their attention. Music therapy becomes an important therapeutic process within a multidisciplinary intervention program that improves thea areas of, imitation, attention, memory and communication, facilitating an appropriate framework for the emotional and social development.

The activity is carried out by the music therapist and volunteers. It will be held once a week with one-hour sessions. The activity takes place last

year in the delegations of Valencia, Valladolid, expanding this year to delegations of Castellon, Badajoz and Madrid.

Valladolid

Badajoz

Valencia

Castellón

Research: "Dynamic eye tracker communicator "

This is a Pilot Study-Research Project whose aim is to find out if our girls can interact with the Communicator. In the Association, we know that Visual Communication (ability to direct the eye) is a well accepted method by the Rett girls and has shown to be adequate to facilitate and encourage interaction with the environment. Wesecky (1986) claims that although difficult, is not impossible for these girls to learn new skills.

Tobbi communicator, has been acquired by the AESR, thanks to a grant from the Caixabank, of Valencia, who showed very enthusiastic with the project and did not hesitate to make it possible. We want to thank all the girls and their families !!!

Methodology: The communicator will rotate every three months by all AESR delegations for an experimental trial of all the girls. BJ technical adaptations will train our communication professionals, and guide them through the process and collect their results.

The project has begun in Valladolid, involving three girls experience carried out by the communications professionals at school.

Genetic research

The Association maintains this year research collaboration with the Foundation INSTITUT D'BELLVITGE Biomedical Research (IDIBELL), recognized and classified as beneficial type of care, with registration number 459, resolution adopted by the Ministry of Justice of the Generalitat de Catalunya.The donation is intended for the Research Program "preclinical trials in mouse models of Rett syndrome: GSK3 mediated inflammation" . The foundation will inform AESR of its results at the expiring date to enable to consider the possibility of future donations to it.

TARGETED TO SOCIETY:

Training

Sensitization: Visibility SR:

What we aim with this activity is to participate in sporting, cultural, educational and health activities. These activities are organized by the Association and other groups are invited to participate in the event by exposing an information stant,in order to inform about RS,and also to have fun, and promote social inclusion.

In sporting events

27 February. "The Center for Sports and Leisure Covaresa - CDO XXI Century - located in Valladolid, organizes a Cycle Master Class in Solidarity with the Spanish Rett Syndrome Association on the occasion of the celebration of World Rare Disease Day to raise awareness and sensitize the existence of these diseases ,including RS.

Result: 80 people Participation

The Spanish Rett Syndrome Association helps Josele (who runs marathon races with his daughter all around the world) to “ push the wheel chair”. On March 22, the ninth edition of the Mountain Marathon was held in Borriol (Castellón).

The Sport Club Atheletic Handball at Valladolid, in their line of collaboration with other organizations, on Saturday April 11 supporting the visibility of Rett syndrome.,

Result: about 100 people Assistance

38th Asprona March at Valladolid.

On Saturday 9th of May we enjoyed a great day of March, 18 kilometer route of solidarity. More than 3,000 walkers made the tour a great day. We also wanted to be there as Spanish Rett Syndrome Association and make visible the disease.

The AESR was one of the entities selected to attend a major sporting event of the Pre-Olympic Women's Hockey team, held in Valencia on June 13th.

HOCKEY
WORLD LEAGUE
Semi-Final

Valencia
Hockey Valencia Stadium
Virgen del Carmen- Beteró
10.06.2015 - 21.06.2015

Women

Argentina	Spain
China	Uruguay
England	USA
Germany	Team 9 Tbc
South Africa	Team 10 Tbc

MAKE YOUR MARK

fih.ch

Also a wonderful opportunity to continue uniting our Rett families through coexistence and thus reminding, parents and children, that they are not alone and that they have the full support of the Spanish Association of Rett syndrome.

It was a very intimate evening, where our children enjoyed a great party, with balloons, music and even a visit from Shrek !!

96041959.

Local Festivity in Mucientes (Valladolid). August. Maria's family put a stand to sell the shirts for AESR and sold a lot. In the picture, the Zumba group “ALL for Rett syndrome” !!!!

Cultural events

El 17 de Enero, **Arganda del Rey Madrid**

On Thursday 5th of March we were present at the ceremony held in the Senate (Madrid) for the International Rare Disease Day. Following the entry of the Health Minister, the President of the Senate and the Queen of Spain proceeded to the delivery of FEDER awards.

“VERSOS Y RELATOS CON RETTO” (various authors)

A book of Poems and stories was presented in Madrid, Valladolid and Toledo to the benefit of AESR


BIBLIOTECA de Castilla-La Mancha
Síndrome de RETT
ASOCIACIÓN ESPAÑOLA www.rett.es
editorial SELEER
HOSPITAL NACIONAL DE PARAPLÉJICOS Toledo

VERSOS Y RELATOS CON RETTO
Antología de autores

La Editorial Seleer, tiene el placer de invitarles a la presentación del libro solidario **“Versos y Relatos con Retto”**, cuyos beneficios serán destinados a la investigación del Síndrome de Rett.

El acto tendrá lugar el miércoles 22 de abril a las 17:30 en el salón de actos del Hospital Nacional de Parapléjicos.

Se contará con la presencia de: Dr. Verdú (Neuropediatra), Javier Rodriguez (Investigador), Lorena Sampedro (Autora y representante de la editorial) y Alejandra Redondo (moderadora).

Con la colaboración de: la Payasa Tomasa, que hará las delicias de los niños, y del Restaurante Jardín-Aldebarán (www.retaurantealdebarantoledo.com)

Participation, on Friday 19th of June , this time organized by FEDER and the San Pablo CEU University of Valencia in the Corte Inglés Store, , in the Social Visibility Campaign "Green dog goes outside."

Elías Durán, Pepa Crespo, Isabel Cosme, Pepe Martínez, Almudena Amaya, Pau Pèrez Rico y Pilar Paricio. LEVANTE-EMV

El perro verde sale a la calle

▶ La Federación de Enfermedades Raras pone en marcha una campaña de visibilidad social

PILAR G. DEL BURGO VALENCIA

■ Los consideran raros aunque no lo son. Solo tienen una enfermedad de baja o muy baja prevalencia y por eso son considerados «extraños», «infrecuentes», «excepcionales», «diferentes»... y porque la mayor parte de las 8.000 enfermedades tipificadas como «raras» que afectan a tres millones de ciudadanos en España no tienen tratamiento ni cura.

Son demasiado pocos para que las multinacionales inviertan en nuevas terapias y fármacos. Demasiados pocos para que a los cuentas. Ese es el drama. Que al ser pocos, es muy fácil cerrar los ojos o esquivar la mirada para no verlos. Por eso, la Federación de Enfermedades Raras, al Universidad CEU Cardenal Herrera y la Confederación de Empresarios del Comercio Valenciano (Cecoval) han arbitrado la campaña del «perro verde» para hacer visible lo que se esconde o se quiere ignorar.

El logo de la iniciativa ha salido de la mano del dibujante Javier Mariscal.

Más de doscientos comercios de la ciudad de Valencia y los centros comerciales más relevantes, así como el Bioparc y el colegio de Veterinarios se han sumado a la propuesta que fue presentada ayer en Valencia por el vicedecano de Publicidad y relaciones Públicas de la CEU-UCH, Pepe Martínez, la delegada de Feder en la C. Valenciana, Almudena Amaya, la presidenta de Cecoval, Isabel Cosme y el director regional de Comunicación y Relaciones Externas de El Corte Inglés, Pau Pérez Rico.

Más de doscientos comercios colocarán muñecos caninos pintados de verde para llamar la atención de los ciudadanos

El «perro verde» se exhibirá de diferentes modos en comercios, centros culturales y puntos estratégicos de la ciudad. Los organizadores van a hacer lacitos para colgarlos de las correas de los perros, según anunció Martínez que señaló que los «perros corpóreos» que se expondrán en las tiendas los han realizado artistas falleros y estudiantes de Bellas Artes. El único objetivo es llamar la atención. Raros pero visibles.

afirma que el
vapor del cigarr
electrónico es
casi inocuo

▶ El informe elaborado po
departamento de Químic
Analítica es el primero qu
se realiza en España

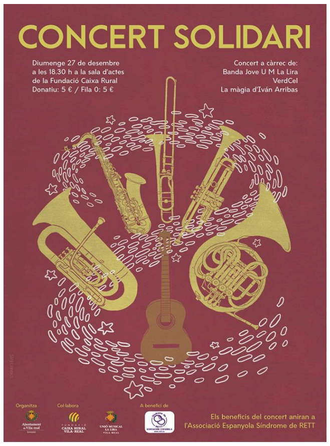
EFE VALENCIA

■ La incidencia del vapor del
rrillo electrónico en ambient
rrados «es nula», ya que la pr
cia de nicotina en el mismo e
inexistente» y su prevalenci
mera», porque se diluye rá
mente en el ambiente. Así c
ye un estudio del Grupo de
tigación «Soluciones e Inno
en Química Analítica» de l
versitat de València, el prim
España que compara el hu
cigarrillo convencional co
por de los cigarrillos electr

El estudio, dirigido por e
drático de Química analíti
UV, Miguel de la Guardia, c
que desde «el punto de vis
tífico, tabaco y cigarrillo ele
co son productos compl
diferentes».

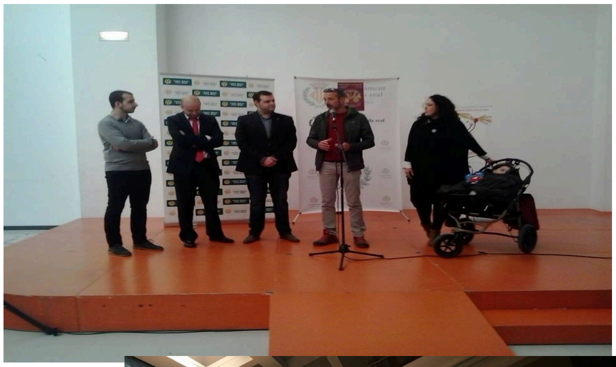
«La nicotina es el únic
diente común entre cigarril
trónicos y tabaco, pero su
cia en el vapor exhalado e
a un 700 % menor que en e
y su prevalencia en el aire e
asegura el experto. Así,
emitido por los cigarrillos
nicos contiene unos 95 n

Concert solidari” Vila-Real



December 27th. This time the initiative "give visibility to the Rett Syndrome" part of the City of Vila-Real (Castellón), in collaboration with the Fundació Caixa Rural Vila-Real and Unió Musical The Lyre of Vila-Real.

Asistencia 200 personas



drome de Rett.

Grupo 1 Secc

Day Center in Cuéllar (Segovia)

Charity market for the Spanish Rett Syndrome Association, Cuellar days 10,11 and 12 December organized by the Territorial Management of Social Services

Junta de Castilla y León

Servicios Sociales de Castilla y León

Centro de Día de Personas Mayores
GERENCIA TERRITORIAL DE SERVICIOS SOCIALES

XVI RASTRILLO SOLIDARIO

Se complace en invitarle a la inauguración del Rastrillo Solidario, a favor de la ASOCIACIÓN ESPAÑOLA DEL SÍNDROME DE RETT, que tendrá lugar en el Centro de Día de Personas Mayores de Cuéllar

Jueves, 10 diciembre

17,00 horas: **Inauguración del Rastrillo Solidario**

17'45 horas: **Actuación Teatro "La Molinera de Arcos"**
Grupo "Tempus Gaudii"

Sábado, 12 diciembre

18'00 horas: **Actuación Danza (danzas orientales, bollywood,…)**
Asociación Intercultural "Aswat Mundi"

Durante los días de celebración del Rastrillo:
Chocolate Solidario

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FILM "Finish Line"

Finish Line "is a film by Paola García Costas that has been a candidate to the nominations for the 2015 Goya awards.

From the Spanish Rett Syndrome Association we have collaborated in disseminating the premiere of the film in all cities in Spain, where it has been issued in order to give visibility to Rett Syndrome.

VALENCIA 12 NOVIEMBRE
MADRID 13NOV, SÁBADO 14 Y DOMINGO 15
VALLADOLID 17 DE NOVIEMBRE
SANTANDER 19 DE NOVIEMBRE
BARCELONA 23 DE NOVIEMBRE
PETRER, (ALICANTE) 3 DE DICIEMBRE
GRANADA 5 DE DICIEMBRE
BADAJOZ 10 DICIEMBRE

From the Spanish Rett Syndrome Association we have collaborated in disseminating the premiere of the documentary in all cities in Spain, where it has been issued in order to give visibility Rett Syndrome.

In AESR, we thank all the participants in this interesting discussion and we believe it has been useful to communicate to the stafft the needs of families suffering the consequences of Rett syndrome. Thank you very much to all!!

ASSINTANCE TO THE CONFERENCES

- Rett Syndrome Conference. Organized by the San Jose Institute Foundation, promoted by the Biorett Fund and the Rotary Club Madrid Puerta del Sol. Febrero. Madrid.

- 5th Conference on Rett Syndrome, organized by the Catalan Rett Syndrome Association. April . Barcelona.

- Intellectual Disability Day. Diagnosis, monitoring and research. Organized by University of Valencia Foundation Chair Genómicos systems. Valencia. November.

- IX Extremadura Rare Disease Day. Organized by the Federation of Rare Diseases (FEDER). Badajoz. November,

-4th European Congress of Rett syndrome. Organized by the Italian Rett Syndrome Association in collaboration with the European CSR. November. Rome.

Members of the European Association of Rett syndrome that attended the European Congress

AIMED AT THE ENTITY

Assemblies

There have been a Assembly on May 30, coinciding the Family Meeting in Madrid.

Boards

They are made on a quarterly basis, resulting throughout the year a total of 4 presential meetings. Have taken place in different parts of Spain still you are in Valencia, Madrid,. It AESR issues and coordination of the activities carried out are discussed. Note that because of the geographical dispersion, besides the physical meetings, the group Board of Directors along with the social worker maintained continuously by meeting points, email and WhatsApp.

Meeting with Management

-Delegation Of Badajoz-(October 16) Health and Social Policy Ministry. Chief of Community Involvement in Health Planning Directorate, Training and Health Quality.

RETT meetings with groups and other entities related to disability.

- CERMI DE EXTREMADURA
- FEDER NACIONAL
- FEDER COMUNIDAD VALENCIANA
- FEDER MADRID
- GRUPOS EMERGENTES DE RETT.

Administration

- Entry in the General Data Protection Registry. Law 15/1999

- Treasury: charging fees, payment receipts, invoices, auditing, other banking matters.

- Administrative tasks such as correspondence, creating dossiers and files etc.

- Database Update: Record high for partners and new partners.

- Study Grants and tracking both government and private entities.

-CHRISTMAS CARDS

- IDENTIFICATION CARDS FOR GIRLS

- CAR STICKERS

RESOURCES

HUMAN

280 Partners of law
25 employees Partners
10 volunteers
Professionals (Social Worker)

MATERIAL

- A 100m2 real estate, divided in two rooms, one for administrative tasks and coordination and anotherone for individual or group interventions.

- Computer and audiovisual equipment, consumables, communication material, educational material, internet and social networks.

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DATA ANALYSIS AND INTERPRETATION

All data expressed in this report conclude in the fact that the Spanish Association of Rett syndrome, begins to assume an important task in society, as well as an active and dynamic role in Family support such as the improvement of visibility of the RS, Training, Rehabilitation, Research and Social Inclusion

The most important aims in AESR are promoting awareness of Rett syndrome, Research, and mainly sensitize on the difficulty for families to live with the consequences of this disease, hence the concern in finding resources and services to ensure the improvement of the the quality of life of children and their families.

ACKNOWLEDGEMENTS

The Spanish Association of Rett syndrome, is grateful to:

- Federation for Rare Diseases (FEDER)
- MINISTRY OF HEALTH AND SOCIAL CARE POLICY OF EXTREMADURA
- Ministry of health. VALENCIAN
- MINISTRY OF EQUALITY POLICIES I INCLUSIVE. GVA
- THE CAIXA
- AIRBUS S.L.
- BROTHERS GALIANO
- State Reference Center for Rare Diseases (CREER)
- INSTITUTE IDIBELL
- SAN JUAN DE DIOS FOUNDATION
- CIRCUS Piruleto ASSOCIATION
- Carrefour Foundation
- And in general all those public institutions, associations and companies who gave us all the means to "be visible".

And especially to the Rett families and volunteers without them, all the work outlined in this report would not have been possible.

AESR JANUARY 2016