

Catalonia Rett Syndrome Association

Pedro Rocha



Catalonian Rett Syndrome Association

A

Hospital attention

- Expert centers
- CATSALUT (Catalonia healthcare)

G

Member services

- Psychologist
- Multi-sensorial room
- Social worker
- Association family get together (yearly)

F

Relationship with other organizations

- Rett Syndrome Europe
- Feder, Nexe, Esclat
- GENCAT

B

Day centers & Residence

- GENCAT agreement
- Signed day centers
- Relationship with Catalonia social care

D

Investigation

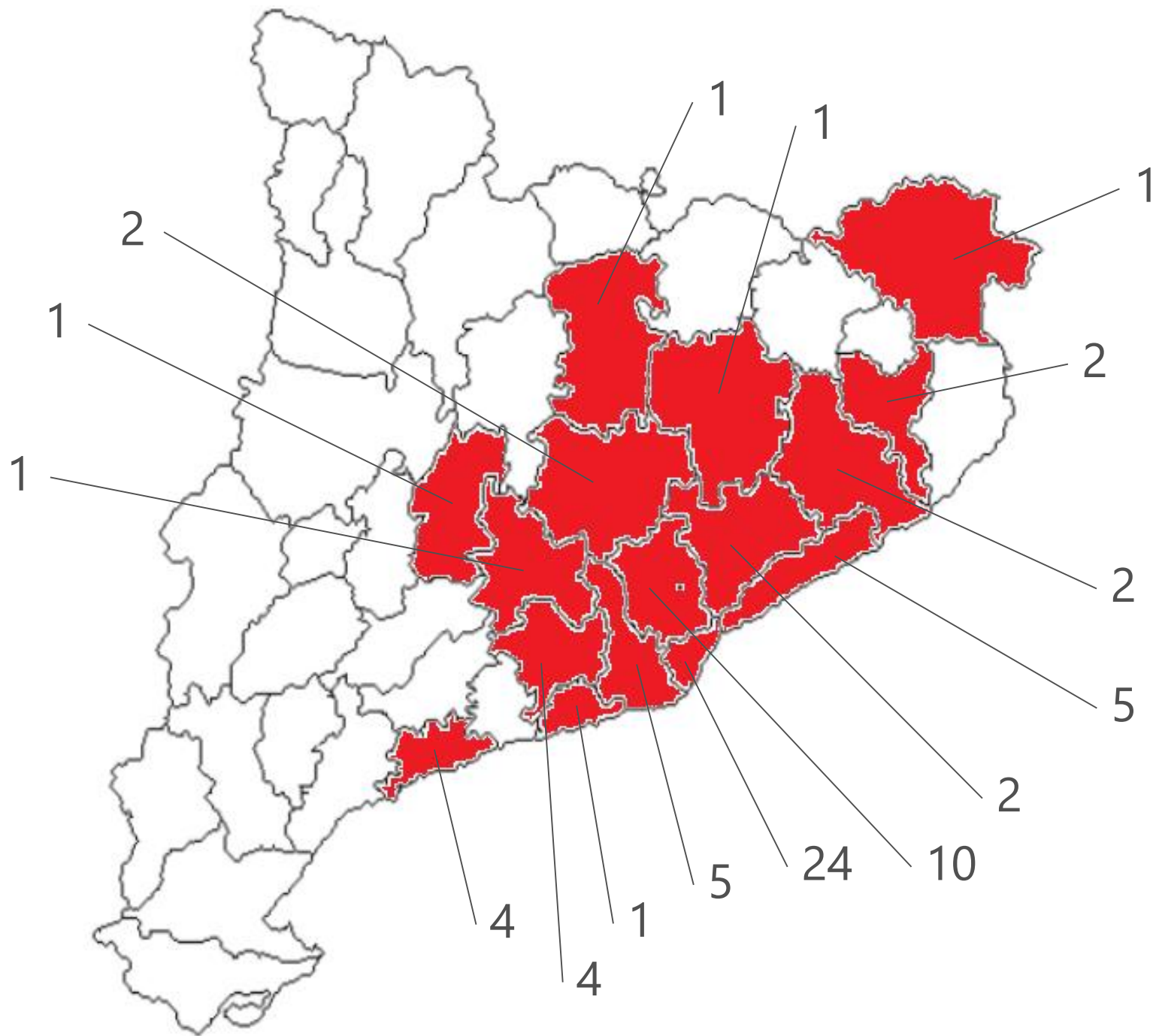
- Support
- Collaboration with hospitals
- Follow-up meetings

E

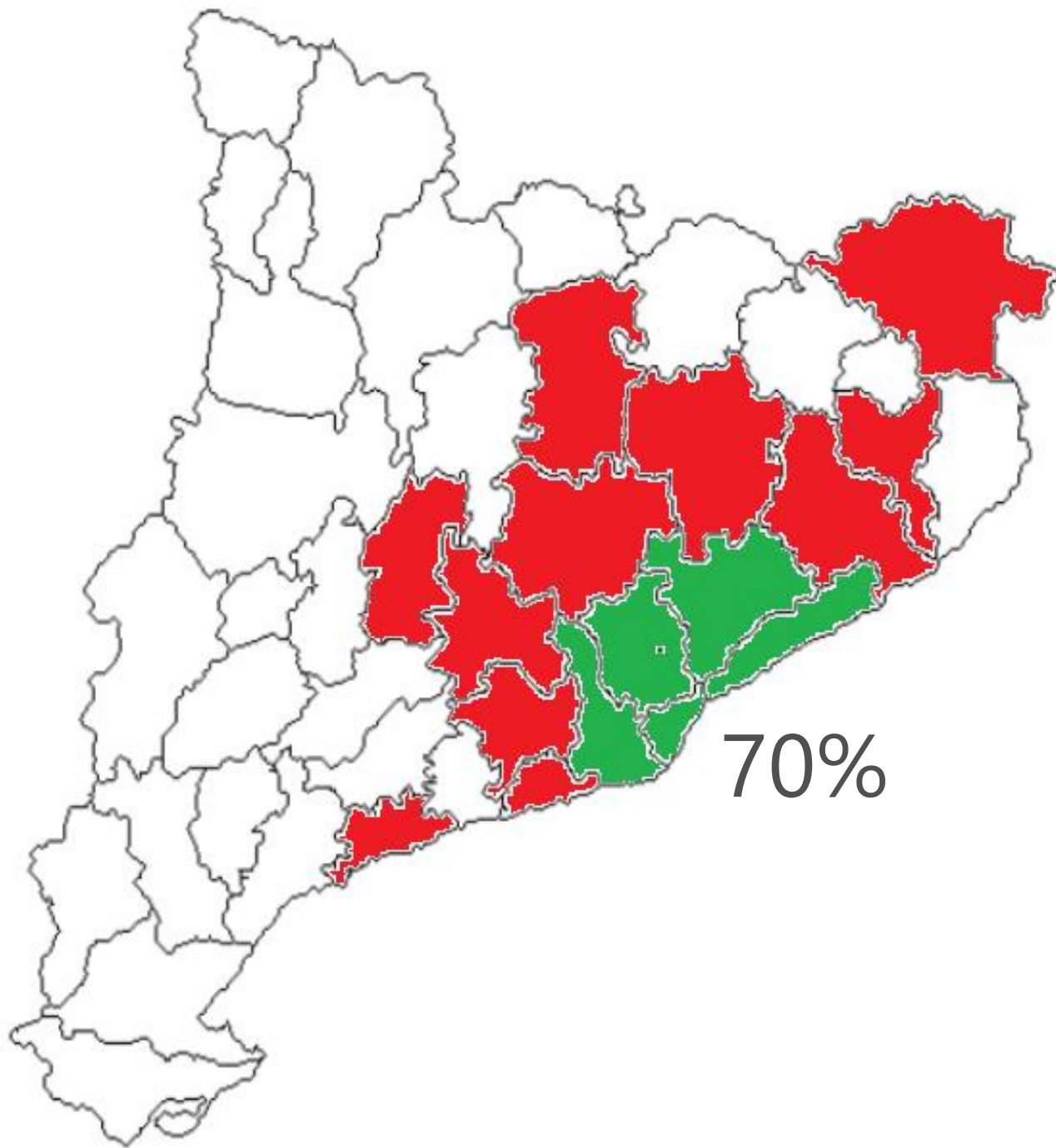
Communication

- Rett day 2014
- New web site & facebook
- Yearly magazine





Area	Rett census
Alt Emporda	1
Alt Penedés	4
Anoia	1
Bages	2
Baix Llobregat	5
Barcelonés	24
Bergueda	1
Garraf	1
Gironés	2
La Selva	2
Maresme	5
Osona	1
Segarra	1
Tarragonés	4
Vallés Occidental	10
Vallés Oriental	2
Total	66



70%

Area	Rett census
Alt Emporda	1
Alt Penedés	4
Anoia	1
Bages	2
Baix Llobregat	5
Barcelonés	24
Bergueda	1
Garraf	1
Gironés	2
La Selva	2
Maresme	5
Osona	1
Segarra	1
Tarragonés	4
Vallés Occidental	10
Vallés Oriental	2
Total	66

Rett expert center

Rett expert center



Children's hospital (public)

+100 Rett children are seen every year
(from month-age to 18 years old)

Reference in Spain for Rett Syndrome



Rett genetic diagnosis

Genetic laboratory with several years of experience in Rett Syndrome diagnosis

Receive Rett diagnosis requests from all Spanish territory

The Rett diagnosis is carried out in the hospital genetic laboratory, using a NGS that covers MECP2, CDKL5, FOXP1 and also other genes (with similarities to Rett like Pitt-Hopking)



Hospital specialists

- Specialisms present in the Hospital:
 - Neurology
 - Pediatrics
 - Physical medicine and rehabilitation
 - Gastroenterology
 - Endocrinology
 - Nutrition, swallow therapy
 - Orthopedic surgery
 - Medical genetics and genetic counselling

Investigation & other activities

- Ongoing activities related with Rett Syndrome:
 - Exome sequencing for discovery of candidate genes in Rett and Rett variants
 - Genetic study of RNA in CSF (Cerebrospinal fluid)
 - Cellular model to study the disequilibrium between excitatory/inhibitory synapses in different Rett mutations
 - Create an informational DVD for families that have a new Rett Syndrome diagnosis
- Tight collaboration with other centers
 - IDIBELL lead by Manuel Esteller
 - University of Barcelona, Bellvitge
 - Instituto de Medicina Molecular de la Universidad de Lisboa
 - Stanford University, USA.

Center funding



The hospital is financed by the public healthcare

Also has a private clinic for patients from outside Catalonia

Sant Joan de Déu Foundation participates in gathering funding for investigation

In 2012, 141 research projects were conducted, 130 of which received financing from Spanish entities and 11 from international entities

Next steps

- Select an expert center for Rett ladies (+18 years)
- Hospital Vall d'Hebron (Barcelona) is currently the main choice for +18 ladies

Rett day centers

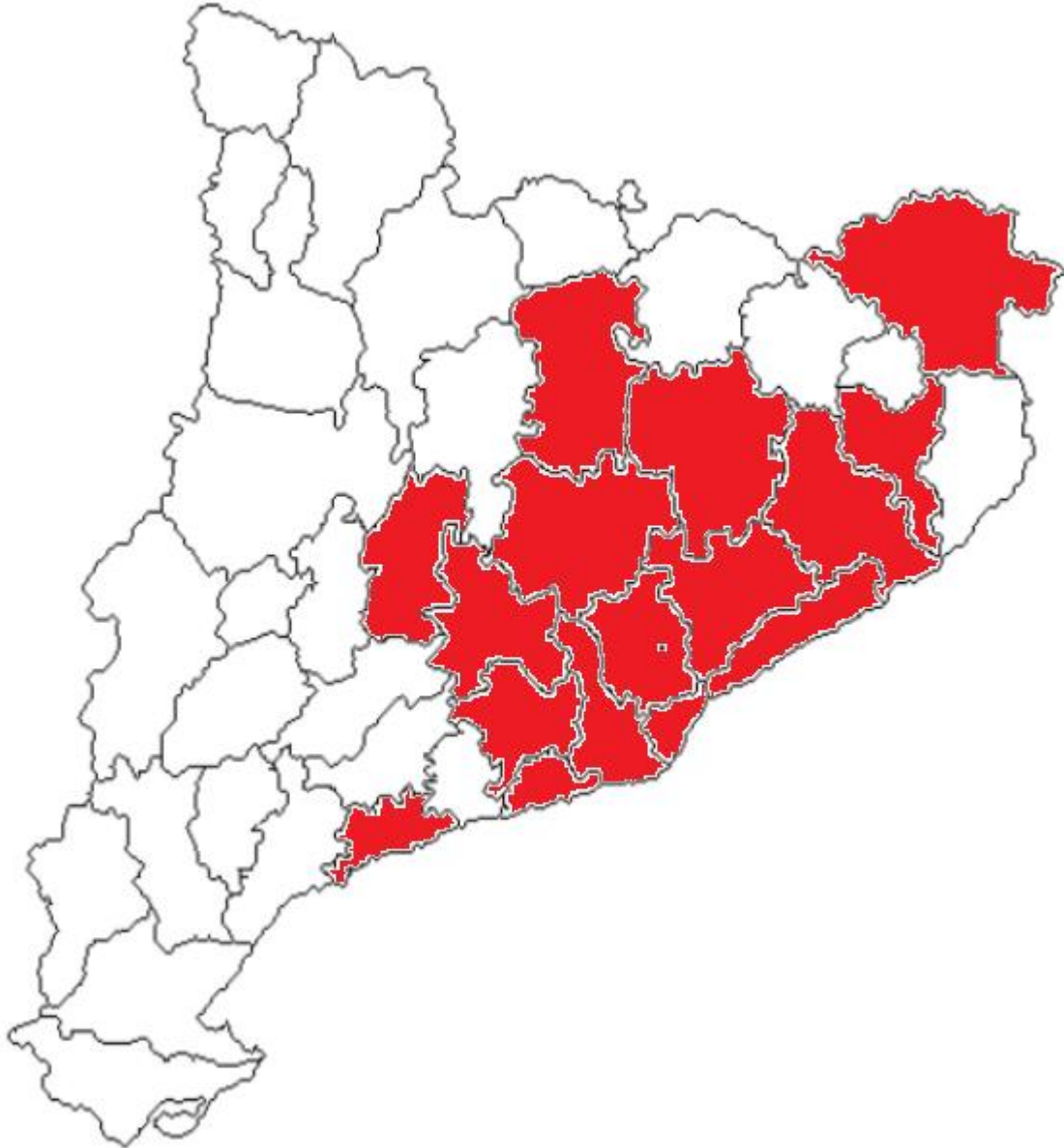
Centers for Rett ladies

- There are no dedicated centers to take care of Rett aged +18
- School finishes at 18, although in special cases can be extended up to 20-21 years old
- In Rett Association signed an agreement with Catalonia Social Services
 - List of centers approved by the Rett Syndrome Association
 - Priority in case there is no available vacancy
- ACSR worked to identify which centers were more suitable (for both day center and residence)

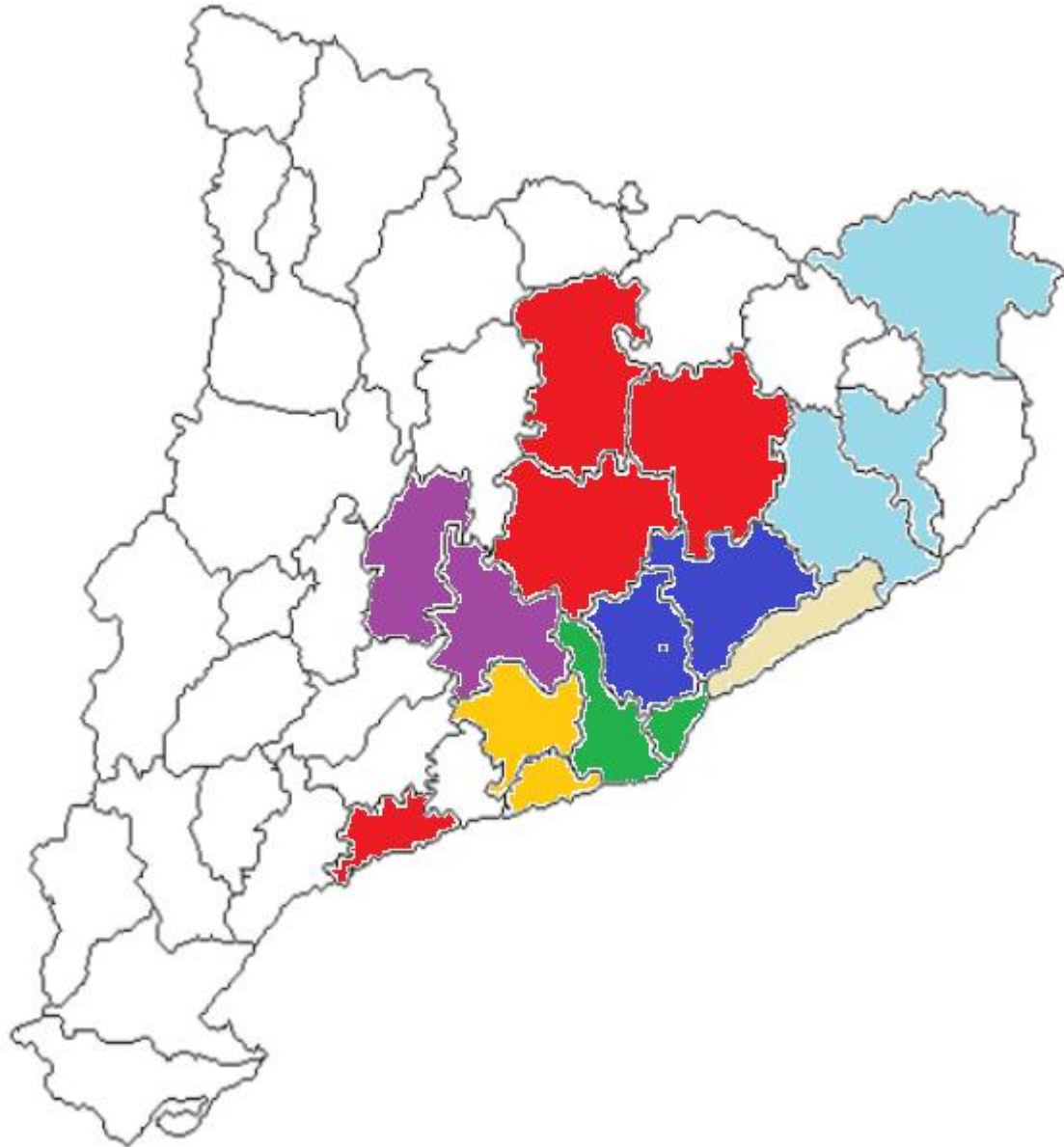
Process to assign a Rett Center


- Every year, the association works with the families to present the day center that applies based on location
- The association acts as the facilitator of the new admission process
- Follow-up to confirm correct integration

Rett center distribution



Rett center distribution



-  Fundación Esclat
-  Fundación Atendis
-  Fundación Delta/Espiga
-  Centro Joan Rius
-  Fundación Aurea
-  Fundación Maresme
-  (no assigned center yet)

Next steps

- New centers identified

- | | |
|--------------------|--------------------------|
| • Tarragonés | Associació La Muntanyeta |
| • Lleida | Associació AREMI |
| • Bages – Bergueda | Associació AMPANS |

- ACSR will visit the centers to assess if these are suitable for Rett Syndrome ladies
- Include these centers in the program or identify new centers
- Continuous follow-up with Catalonia Social Services

