

Asociación Española del Síndrome de  
**Beckwith-Wiedemann**



**BREMEN 2015**

# ASEBEWI?



- Founded in 2007
- Encourage Knowledge of BWS
- 41 families in Spain
- A lot of contacts from South America in our forum

# BWS



- Beckwith-Wiedemann syndrome (BWS) is an overgrowth syndrome associated with an increased risk of pediatric tumors. The underlying molecular abnormalities may be genetic (CDKN1C mutations or 11p15 paternal uniparental isodisomy, pUPD) or epigenetic (imprinting center region 1, ICR1, gain of methylation, ICR1 GOM, or ICR2 loss of methylation, ICR2 LOM).
  - Nowadays there aren't different protocols for each kind of BWS
  - Main problems
    - Macroglossia
    - Abdominal wall defect
    - Exomphalos
    - Organomegaly
    - Hemihyperplasia
    - Hypoglycemia
    - Ear abnormalities
    - Facial nevi
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# What are we doing in ASEBEWI?



- Support, advice, experience sharing to the families
- Look for doctors and reference people
- Two meeting/year:
  - Ordinary meeting in Madrid
  - Trip for a weekend
- Colaboration with FEDER (rare diseases association)
- New web site [www.asebewi.org](http://www.asebewi.org)
- New facebook group. [www.facebook/asebewi](http://www.facebook/asebewi) (Closed)
- Radio programs, newspaper...
- **Encourage to medical community to update a clinical guide**
- We will have the first Spanish convention in Hospital La Paz in Madrid (30th of May)

# What are we doing in ASEBEWI?



## New ideas

- Develop some flyers/papers
- Implementing a coordinator for Rare (BWS) diseases ...
- Data base with doctors (and speciality)
- Connecting with other European/American BWS Association
- Look for international experts
- Compare different protocols/techniques/surgeries
- Promote new members to action

# What are we doing in ASEBEWI?



## Problems...

- Few specialist in Spain (little knowledge about the disease)
- Look for new resources (money)
- Look for a medical advisor. (Geneticist, endocrine, speech therapy,...?)
- Very different views about protocols, surgeries, etc.

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On behalf of Cloe&Asebewi Thank you everybody