



A.I.S.EA Family Meeting
Rome, 26 – 27 March 2011



AESHA Presentation

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Outline

- ▶ Introduction
 - ▶ Members
 - ▶ Goals
 - ▶ Achievements
 - ▶ Projects
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The AESHA association


- ▶ Created in 2004
- ▶ Member of FEDER: Spanish Federation of Rare Diseases (since 2005)
- ▶ Member of ENRAH (since 2005)
- ▶ Scientific Committee:
 - Dr. Jaume Campistol (Hospital St. Joan de Déu, Barcelona)
 - Dr. Carme Fons (Hospital St. Joan de Déu, Barcelona)
 - Dr. Ramón Velázquez (Hospital Infantil La Paz, Madrid)

Members

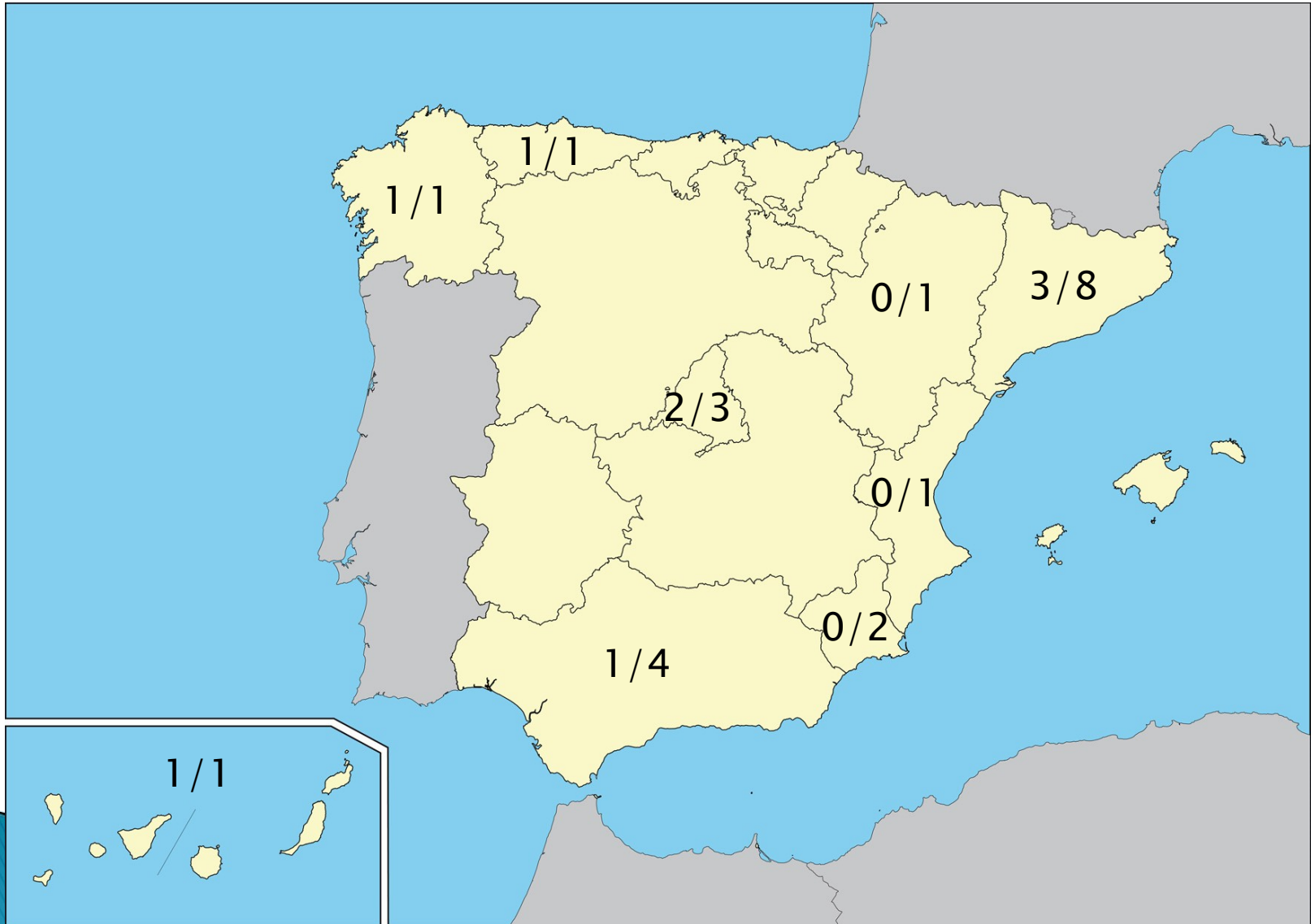
- ▶ Affectation of AHC: 1 case per 1.000.000
 - ▶ Spanish Population: ~ 43.000.000

 - ▶ 22 known cases
 - ~ 50% non identified cases

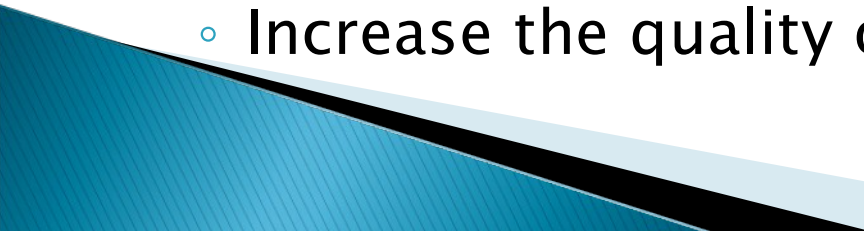
 - ▶ 9 cases in our association
 - ~ 25% belong to the association

 - ▶ 2 cases having familiar hemiplegic migraine
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Geographic distribution



Goals

- ▶ Spread the disease knowledge
 - Society
 - Medical and scientific community
 - ▶ Bring members together
 - Truthful and updated information
 - Advise
 - Knowledge exchange
 - ▶ Support the research
 - Increase the knowledge on AHC
 - Increase the quality of life
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Achievements

- ▶ Medical Guide at UdG
- ▶ ENRAH
- ▶ nEUroped
- ▶ AHC White Book
(2.000 copies)



National research

- ▶ Spanish DNA Biobank (since 2008)
 - *C. Fons, L. Martorell, J. Campistol – Molecular Genetics Department HSJD*
 - 16 patients and direct relatives
- ▶ Metabolic study on AHC
 - *C. Fons, C. Villar, J. Campistol, E. Panagiotakai, A. Arzimanoglou*
 - European series of 157 patients
 - To appear
- ▶ GLUT1 Study in Spanish series
 - *J. Campistol – HSJD*
 - Genetics: 6 negatives
 - Lumbar Puncture: 2 negatives + 2 positives

Current projects

▶ Scientific

- Participation in I.B.AHC (A.I.S.EA)
- Participation in the Study of Paroxistical Events (A.I.S.EA)

▶ Social

- National Registry of Rare Diseases (FEDER)
- Multilanguage online community on AHC (EURORDIS)

Any question?



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<http://www.aesha.org>