InterRett
International database

Alison Anderson
World Rett Congress
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Overview

- What is InterRett
- Objectives
- Achieved to date
- Future challenges
• International project to collect information about individuals diagnosed with Rett syndrome

• Funded by IRSF - International Rett Syndrome Foundation

• Managed by the Australian Rett syndrome team based in WA

• Commenced in 2002
Objectives

1. Provide a large sample of cases for analysis
2. Provide an innovative and efficient mechanism to disseminate information
3. Encourage collaboration around the world
Objective 1

1. Provide a large sample of cases for analysis – why?

To increase statistical power

n=74

n=836
Provide a large sample

- Studies are limited by the number of cases available on a national level

- Even more so if they are investigating specific mutations or characteristics

- Solution is to pool data from multiple countries
Objective 1

1. Provide a large sample of cases for analysis – how?

   Invite families to participate
     • Website
     • Family associations

   Family provide Consent
   Complete questionnaire (online/paper-based)

   Invite a clinician to provide clinical information
Clinicians who have multiple patients can submit de-identified information on all their patients.

### Contributing clinicians

The following clinicians have contributed information on one or more of their patients to the InterRett database.

<table>
<thead>
<tr>
<th>By Surname</th>
<th>By Country</th>
<th>Number per country</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Dr Aaron, USA</td>
<td>Dr Di Elasi, ARGENTINA</td>
<td>Australia 8</td>
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<tr>
<td>2. Dr Abenheimer, CANADA</td>
<td>Dr Munt, AUSTRALIA</td>
<td>Canada 20</td>
</tr>
<tr>
<td>3. Dr Abunto, USA</td>
<td>Dr Sheffield, AUSTRALIA</td>
<td>New Zealand 5</td>
</tr>
<tr>
<td>4. Dr Alper, USA</td>
<td>Dr Churchyard, AUSTRALIA</td>
<td>UK 40</td>
</tr>
<tr>
<td>5. Dr Anderson, USA</td>
<td>Dr Crowhurst, AUSTRALIA</td>
<td>USA 145</td>
</tr>
<tr>
<td>6. Dr Appleton, AUSTRALIA</td>
<td>Dr Delatycki, AUSTRALIA</td>
<td>Other 24</td>
</tr>
<tr>
<td>7. Dr Ashwal, USA</td>
<td>Dr Ellaway, AUSTRALIA</td>
<td></td>
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<tr>
<td>8. Dr Atkin, USA</td>
<td>Dr Balkie, AUSTRALIA</td>
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<tr>
<td>9. Dr Augenstein, USA</td>
<td>Dr Appleton, AUSTRALIA</td>
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</tbody>
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Total = 242
Objective 1 Results

Family information (individual families) Total (623)

- Both family and clinical information: 346
- Clinical Information (Total 429):
  - From clinicians with one or a few patients: 277
  - From clinicians with large datasets: 152

Clinician data only: 152
Family & clinician: 277
Spanish: 354
French: 231
Australia: 331
Total: 1,691

Spanish data (Dr Pineda’s questionnaire)
French data Coded (Adaptation of InterRett questionnaire)
Australian data (Australian questionnaire)
Other = countries with 10 or less participants:

Argentina, Austria, Belgium, Bolivia, Brazil, Chile, Colombia, Denmark, Germany, Honduras, Hungary, India, Iran, Ireland, Italy, Japan, Malta, Mexico, Netherlands, New Zealand, Norway, Peru, Portugal, South Africa, Sweden, Switzerland, Taiwan, Turkey, United Arab Emirates, Uruguay
**Mutation status**

**Mutation test**

<table>
<thead>
<tr>
<th></th>
<th>Yes</th>
<th>No</th>
<th>Unknown</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Positive</td>
<td>1010</td>
<td>316</td>
<td>157</td>
<td>1483</td>
</tr>
<tr>
<td>Negative</td>
<td>1483</td>
<td>155</td>
<td>53</td>
<td>1691</td>
</tr>
</tbody>
</table>

652 with common mutations
2. Provide an innovative and efficient mechanism to disseminate information – why?

Increase the clinical understanding of Rett syndrome

Maximise the use of information collected

Take advantage of information technology
Welcome to the InterRett Output Database

This is a searchable database which allows you to create graphs based on the information provided in the InterRett family questionnaires from families around the world.

Create a graph of one variable

OR Compare two variables
Objective 2

Publications


Objective 3

3. Encourage collaboration around the world

- In September 2007 a China-Australian workshop in Beijing (~ 80 families)
- Funded by a travel grant awarded to InterRett from the Australia-China Fund for Science & Technology
- Build on their existing relationship with Dr Wu and Dr Bao
Objective 3

- **Scoliosis guidelines** (Clinical management of scoliosis workshop J. Downes)
  - Using the Internet to build consensus
  - Meaningful results from a simple infrastructure
  - Approach can be applied to other research objectives
• Internet can successfully be used for collecting information and building consensus

• Data sharing makes sense
  – Improved statistical power
  – Avoids duplication of effort
  – Reduces burden on families
  – Reduces cost to funding bodies
• Challenges
  – Need to protect patient privacy
    • Informed consent
  – Intellectual property rights of researchers
    • Collaborate but remain competitive
  – Lack of standardisation
  – Cottage industry approach
The way forward

• We need consensus on how to proceed
• Establish common protocols (measurement Ami Bebbington’s talk)
• Take stock
  – Why and when we should pool data
  – Identify all the data sources available
  – What can be shared and what can’t
  – Avoid duplications
Thanks!

Families & Clinicians
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Mercedes Pineda, Nadia Bahi-Buisson,
Bruria Ben-zeev, Patrick McLeod
Dr Bao, Dr Wu

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IRSF

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