Epilepsy in Rett syndrome
– from a lifetime perspective

Mari Wold Henriksen
MD/Phd-student
Drammen hospital VVHF/University of Oslo
Norway
A national survey on females with Rett syndrome in Norway

Main aims:

• Describe the phenotypic variation in the Norwegian Rett Syndrome population, and the development of clinical features in different phases of life.

• Examine which treatment and habilitation strategies offered to this group
Methods

N≈160

✓ Interview
✓ Clinical examination
✓ Genetic work-up
✓ Review of medical journals

93

Excluded N=2

Classic RTT N=74

Atypical RTT N=10

RTT-like N=7
Epilepsy in Rett Syndrome

- Prevalence 70-90%
- Seizure onset 3-5 years
- Multiple seizure types
- Pattern of remission/relapse?
Pattern of remission/relapse?

• The Rett Syndrome natural History Study
• Prospective cohort
• Up to 8 years of follow-up
• 1205 participants
Aged 8 months-66 years
Epilepsy in Rett syndrome

No seizures

Remission/relapse

Never experienced remission

Ever

During study
- Average duration 1 year
- 7% ≥ five years

- 41% of study population

Tarquinio et al., 2017. Longitudinal course of epilepsy in Rett syndrome and related disorders, Brain 140, 306-318
No seizures

Remissions and relapses

Never remissions

Tarquinio et al.

Henriksen et al.
Epilepsy in Rett Syndrome

- Prevalence 70-90%
- Seizure onset 3-5 years
- Multiple seizure types
- Better with increasing age
- Pattern of remission/relapse?
Aim:
To describe the course and characteristics of epilepsy in adults with Rett syndrome

Better with increasing age?
N≈160

N=93

✓ Interview
✓ Clinical examination
✓ Genetic investigation
✓ Review of medical journals

Excluded N=2

Classic RTT N=74

Atypical RTT N=10

RTT-like N=7
N≈160

Classic RTT N=74
- Interview
- Clinical examination
- Genetic investigation
- Review of medical journals

Atypical RTT N=10

RTT-like N=7

Excluded N=2

Missing data n=2

Epilepsy study N=72
Age

Epilepsy study
N=72

- 1-10 years
- 11-20 years
- 21-30 years
- >30 years
Results
Prevalence of active epilepsy
Results

Prevalence of active epilepsy

- 1-10 years: 7 never seizures, 4 active epilepsy, 2 seizure free last five years
- 11-20 years: 12 never seizures, 10 active epilepsy, 2 seizure free last five years
- 21-30 years: 4 never seizures, 3 active epilepsy, 4 seizure free last five years
- > 30 years: 14 never seizures, 14 active epilepsy
Results

Frequency of seizures

<table>
<thead>
<tr>
<th>Age Range</th>
<th>&lt; monthly</th>
<th>&lt; weekly, ≥ monthly</th>
<th>≥ weekly</th>
</tr>
</thead>
<tbody>
<tr>
<td>11-20 years n=12</td>
<td>6</td>
<td>2</td>
<td>4</td>
</tr>
<tr>
<td>21-30 years n=10</td>
<td>3</td>
<td>5</td>
<td>2</td>
</tr>
<tr>
<td>&gt;30 years n=14</td>
<td>2</td>
<td>7</td>
<td>5</td>
</tr>
</tbody>
</table>
## Results

### Seizure types

<table>
<thead>
<tr>
<th>Age Group</th>
<th>N</th>
<th>Bilateral tonic-clonic seizures N(%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>11-20 years</td>
<td>12</td>
<td>7 (58)</td>
</tr>
<tr>
<td>21-30 years</td>
<td>10</td>
<td>7 (70)</td>
</tr>
<tr>
<td>&gt; 30 years</td>
<td>14</td>
<td>9 (64)</td>
</tr>
<tr>
<td>Women</td>
<td>&gt;30 years</td>
<td></td>
</tr>
</tbody>
</table>
Never seizures

Women

>30 years
Seizure free last 5 years

Women >30 years
Active epilepsy

Women >30 years
Conclusion

• Many will experience a pattern of remissions and relapses

• Epilepsy is still a major concern in many adults with Rett syndrome
Acknowledgements

Coauthors:

- Clinicians:
  - Ola H. Skjeldal
  - Eylert Brodtkorb

- Psychologists:
  - Stephen von Tetzchner
  - Hilde Breck

- Medical geneticist:
  - Benedicte Paus

All girls/women and families participating in this project

The Norwegian Rett syndrome parent association

Vestre Viken Hospital Trust for funding