LANGUAGE DEVELOPMENT
IN CHILDREN WITH CHROMOSOME 14 ABERRATIONS

Laura D’Odorico, Laura Zampini, Paola Zanchi

Department of Psychology, University of Milan-Bicocca
International Association RING14 Onlus, Reggio Emilia

laura.zampini1@unimib.it

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Ring Chromosome 14 Syndrome:

A rare genetic disorder (with 50 cases ever reported) caused by an anomaly in the 14th chromosome, where a rearrangement occurs creating a ring-like formation, generally associated with a partial loss of genetic material.

Linear deletions:

Genetic disorders caused by the loss of a segment of the 14th chromosome.
Most Common Clinical Characteristics

RING CHROMOSOME 14 SYNDROME

• Delays in psychomotor and language development
  • Behavioral problems
  • Autistic traits (stereotyped behavior/qualitative impairment in social interaction/restricted interests)
    • Cerebral morphological anomalies
      (microcephaly/hypoplasia/agenesia of the corpus callosum)
      • Facial dysmorphisms
  • Epilepsy (often early-onset and drug-resistant)
  • Anomalies of the retinal pigment epithelium
In children with chromosome 14 aberrations there is a large phenotypic variability (due to associated pathologies, cerebral morphological anomalies...) and a notable variability in the neuropsychological development (Zollino et al, 2009)

the aim of this study is to describe
the communicative and linguistic abilities
of Italian children with Ring 14 and deletions,
giving particular attention to the influence of three factors:

- the presence or absence of autism
- the presence or absence of epilepsy
- the presence of cerebral morphological anomalies
The language development assessment was administered to 15 children ranging in age from 2 to 16 years with either Ring 14 (n = 6) or chromosome 14 deletions (n = 9).

Analysis of the spontaneous language production while interacting with an adult.

The 8 youngest children (2-7 years old) are longitudinally followed, in order to evaluate the developmental trend of their communicative abilities.
1- Description of the level of language development reached by the participants (analysis of the first session)

2- Exemplificative comparison of some cases, revealing the influence of the considered factors (type of gene anomaly, presence of autism, epilepsy and cerebral morphological anomalies) on language development
## Participants with Chromosome 14 Deletions

<table>
<thead>
<tr>
<th>Name</th>
<th>Age (years.months)</th>
<th>Gender</th>
<th>Genetic Anomaly</th>
<th>Autism</th>
<th>Epilepsy</th>
<th>Morphological Anomalies</th>
<th>Level of Language Development</th>
</tr>
</thead>
<tbody>
<tr>
<td>SI</td>
<td>03:04</td>
<td>F</td>
<td>Deletion</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>Preverbal</td>
</tr>
<tr>
<td>DMR</td>
<td>05:11</td>
<td>F</td>
<td>Deletion</td>
<td>++</td>
<td>-</td>
<td>+</td>
<td>Single-word Development</td>
</tr>
<tr>
<td>DMT</td>
<td>05:11</td>
<td>F</td>
<td>Deletion</td>
<td>++</td>
<td>-</td>
<td>+</td>
<td>Combinatorial Development</td>
</tr>
<tr>
<td>AS</td>
<td>07:00</td>
<td>M</td>
<td>Deletion</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>Complex</td>
</tr>
<tr>
<td>LL</td>
<td>07:08</td>
<td>M</td>
<td>Deletion</td>
<td>-</td>
<td>-</td>
<td>+</td>
<td></td>
</tr>
<tr>
<td>BF</td>
<td>07:08</td>
<td>F</td>
<td>Deletion</td>
<td>-</td>
<td>-</td>
<td>++</td>
<td></td>
</tr>
<tr>
<td>CG</td>
<td>10:03</td>
<td>F</td>
<td>Deletion</td>
<td>+</td>
<td>-</td>
<td>++</td>
<td></td>
</tr>
<tr>
<td>SF</td>
<td>15:00</td>
<td>M</td>
<td>Deletion</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>FE</td>
<td>15:10</td>
<td>F</td>
<td>Deletion</td>
<td>-</td>
<td>+</td>
<td>-</td>
<td></td>
</tr>
</tbody>
</table>
Participants with Ring 14

<table>
<thead>
<tr>
<th>Name</th>
<th>Age (years/months)</th>
<th>Gender</th>
<th>Genetic Anomaly</th>
<th>Autism</th>
<th>Epilepsy</th>
<th>Morphological Anomalies</th>
<th>Level of Language Development</th>
</tr>
</thead>
<tbody>
<tr>
<td>FG</td>
<td>02;08</td>
<td>F</td>
<td>Ring</td>
<td>-</td>
<td>+</td>
<td>+</td>
<td>x</td>
</tr>
<tr>
<td>PR</td>
<td>04;10</td>
<td>M</td>
<td>Ring</td>
<td>+</td>
<td>+</td>
<td>-</td>
<td>x</td>
</tr>
<tr>
<td>RM</td>
<td>08;01</td>
<td>M</td>
<td>Ring</td>
<td>+</td>
<td>++</td>
<td>-</td>
<td>x</td>
</tr>
<tr>
<td>GG</td>
<td>10;07</td>
<td>M</td>
<td>Ring</td>
<td>++</td>
<td>++</td>
<td>-</td>
<td>x</td>
</tr>
<tr>
<td>FS</td>
<td>12;01</td>
<td>F</td>
<td>Ring</td>
<td>-</td>
<td>+</td>
<td>+</td>
<td>x</td>
</tr>
<tr>
<td>RJ</td>
<td>15;03</td>
<td>M</td>
<td>Ring</td>
<td>+</td>
<td>+</td>
<td>++</td>
<td>x</td>
</tr>
</tbody>
</table>
Comparison 1 [FG_Ring – SI_Del]

2 children with different genetic anomalies:

- FG (02;08) F \(\rightarrow\) Ring 14
  (No autism/epilepsy(controlled)/minor cerebral morphological anomalies)

- SI (03;04) F \(\rightarrow\) Deletions
  (No autism/ No epilepsy/ No cerebral morphological anomalies)

<table>
<thead>
<tr>
<th>FG_Ring</th>
<th>SI_Del</th>
</tr>
</thead>
<tbody>
<tr>
<td>No sitting without support</td>
<td>Autonomous deambulation</td>
</tr>
<tr>
<td>Minimal in-hand manipulation skills</td>
<td>Symbolic Play</td>
</tr>
<tr>
<td>Motor Development</td>
<td>Level of Play</td>
</tr>
</tbody>
</table>
Comparison 1 – Language Development

Vocal Production

- FG_Ring: 64 utterances in 20 minutes of interaction
- SI_Del: 63 utterances in 20 minutes of interaction

Gesture production

- FG_Ring: 8 gestures
- SI_Del: 16 gestures

`pointing` `showing` `ritualized_request` `conventional` `iconic`
Comparison 2 [GG_Ring – FS_Ring]

2 children with Ring 14:

• GG (10;07) M → Ring 14
  Autism/ drug resistant epilepsy/ No cerebral morphological anomalies

• FS (12;01) F → Ring 14
  No autism/ controlled epilepsy/ minor cerebral morphological anomalies
  (white matter hypoplasia)

<table>
<thead>
<tr>
<th>Mental Age (CPM)</th>
<th>GG_Ring</th>
<th>FS_Ring</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>04:03</td>
<td>06:00</td>
</tr>
</tbody>
</table>
## Comparison 2 – Language Development

### Types of different words produced in 15 minutes of interaction

<table>
<thead>
<tr>
<th>Type</th>
<th>GG_Ring</th>
<th>FS_Ring</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>46</td>
<td>140</td>
</tr>
</tbody>
</table>

### Mean length of utterance

<table>
<thead>
<tr>
<th>MLU in words</th>
<th>GG_Ring</th>
<th>FS_Ring</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>2</td>
<td>2.71</td>
</tr>
</tbody>
</table>
2 children with chromosome 14 deletions:

- LL (07;08) M → Deletion  
  No autism/ No epilepsy/ minor cerebral morphological anomalies (thinning of the corpus callosum)

- BF (07;08) F → Deletion  
  No autism/ No epilepsy / several cerebral morphological anomalies (hypoplasia of the corpus callosum, opercular deficit)
Comparison 3 – Psychomotor Development

Griffiths Scales (administered at 8 years of chronological age)

LL_Del: m.a. = 03;08  
BF_Del: m.a. = 01;04
Comparison 3 – Language Development

Number of utterances produced in 20 minutes of interaction

- LL_Del: Preverbal utterances
- BF_Del: Verbal utterances

Verbal utterance composition LL_Del
- Single-words
- Utterances without verbs
- Simple sentences
- Complex sentences

Preverbal utterance composition BF_Del
- Preverbal level 1
- Preverbal level 2
Participants’ description:
The linguistic and communicative competences of children with Ring 14 and chromosome 14 deletions are NOT homogeneous

Comparison FG_Ring – SI_Del:
• The language development of children with Ring 14 proves to be generally more impaired, also due to the greater number of associated pathologies (a more critical or drug-resistant epilepsy and more frequent autism)
Conclusions

Comparison GG_Ring – FS_Ring:
• Within the group of children with Ring 14, a notable influence from epileptic seizures and autistic traits is revealed
→ Only FS, who has drug-controlled seizures and no autistic traits, has a good language development

Comparison LL_Del – BF_Del:
• Consistent with other conditions, it is presumable that the cerebral morphological anomalies have an influence on the psychomotor and linguistic development of children with the same type of genetic anomaly
This research has been funded by
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www.ring14.org