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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Genetic Anomalies in the 14th Chromosome

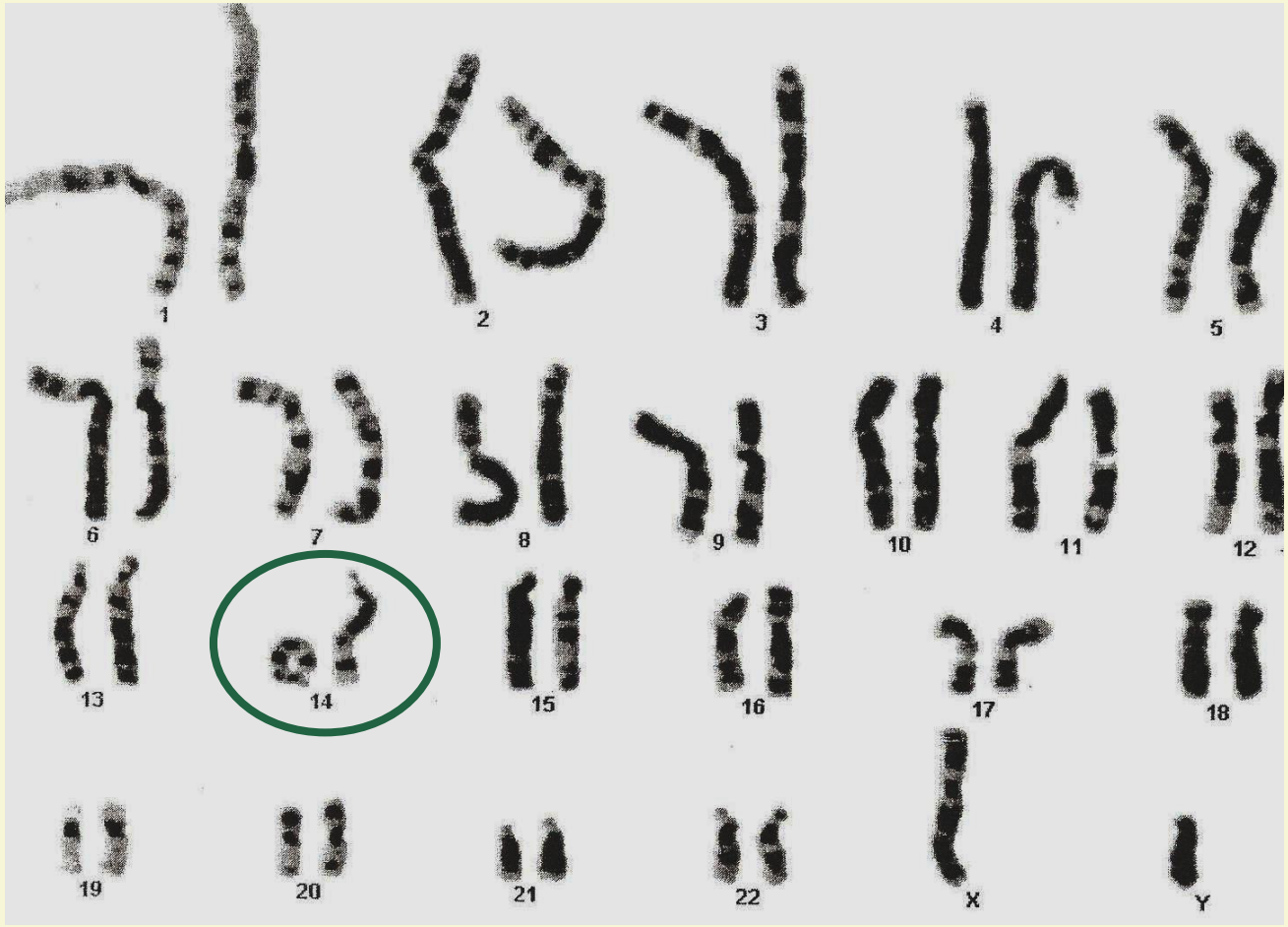
Ring Chromosome 14 Syndrome:

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

Karyotype Ring14



Most Common Clinical Characteristics

RING CHROMOSOME 14 SYNDROME **DELETIONS**

- **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**

Aim of the Project

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**

Participants and Procedure

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

Name	Age (years;months)	Gender	Genetic Anomaly	Autism	Epilepsy	Morphological Anomalies	Level of Language Development			
							Preverbal	Single-word	Combinatorial	Complex
SI	03;04	F	Deleletion	-	-	-	x			
DMR	05;11	F	Deleletion	++	-	+	x			
DMT	05;11	F	Deleletion	++	-	+	x			
AS	07;00	M	Deleletion	-	-	-				x
LL	07;08	M	Deleletion	-	-	+				x
BF	07;08	F	Deleletion	-	-	++		x		
CG	10;03	F	Deleletion	+	-	++	x			
SF	15;00	M	Deleletion	-	-	-		x		
FE	15;10	F	Deleletion	-	+	-				x

Participants with Ring 14

Name	Age (years;months)	Gender	Genetic Anomaly	Autism	Epilepsy	Morphological Anomalies	Level of Language Development			
							Preverbal	Single-word	Combinatorial	Complex
FG	02;08	F	Ring	-	+	+	x			
PR	04;10	M	Ring	+	+	-	x			
RM	08;01	M	Ring	+	++	-		x		
GG	10;07	M	Ring	++	++	-			x	
FS	12;01	F	Ring	-	+	+				x
RJ	15;03	M	Ring	+	+	++			x	

Comparison 1 [FG_Ring – SI_Del]

2 children with different genetic anomalies:

- **FG (02;08) F → Ring 14**

(No autism/epilepsy(controlled)/minor cerebral morphological anomalies)

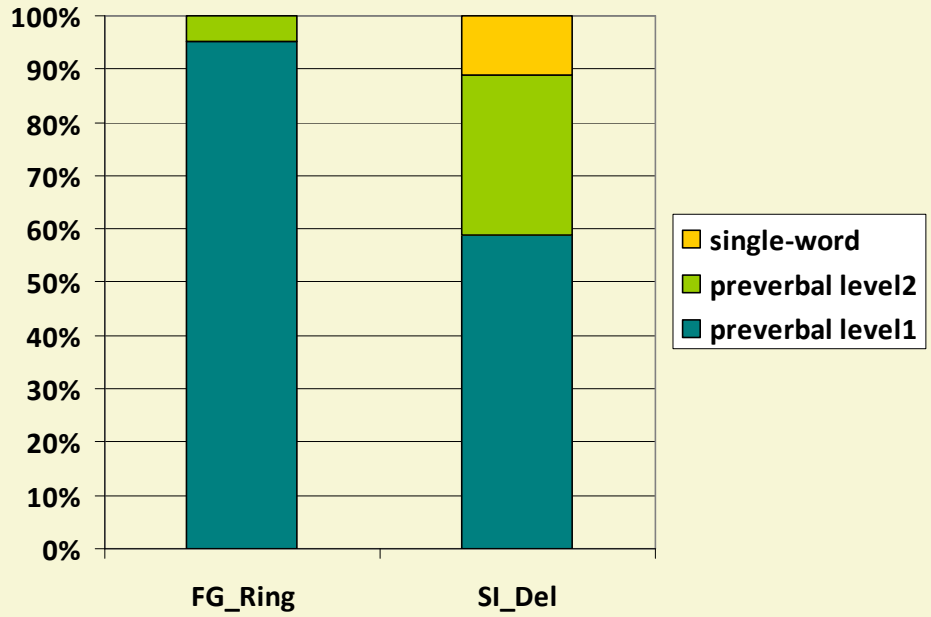
- **SI (03;04) F → Deletions**

(No autism/ No epilepsy/ No cerebral morphological anomalies)

FG_Ring		SI_Del
No sitting without support	Motor Development	Autonomous deambulation
Minimal in-hand manipulation skills	Level of Play	Symbolic Play

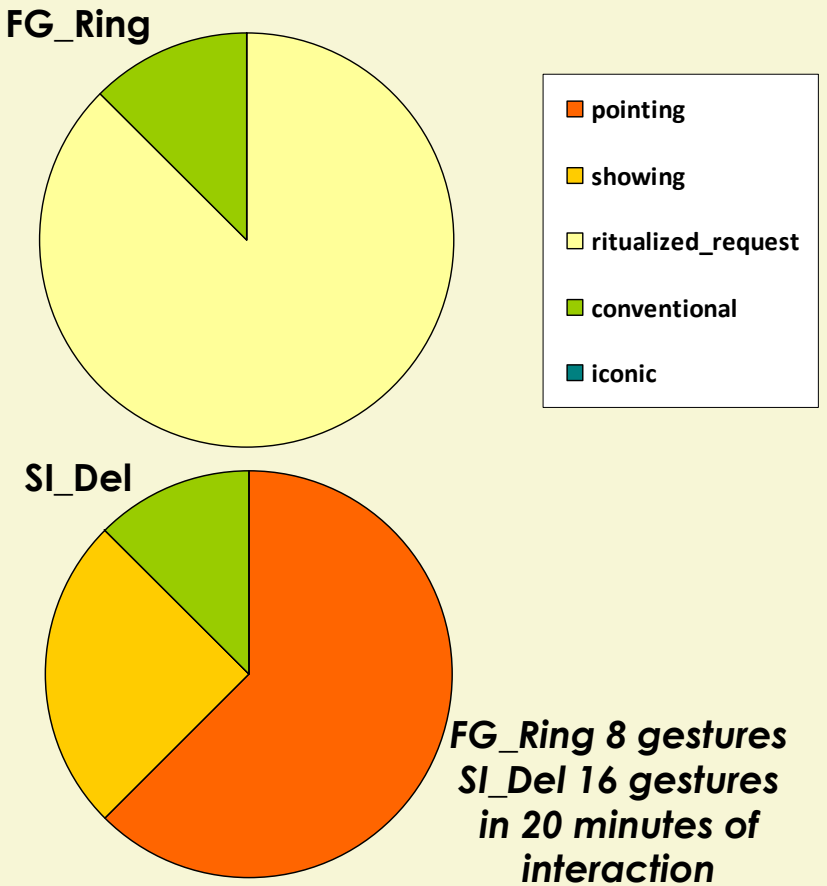
Comparison 1– Language Development

Vocal Production



FG_Ring 64 utterances
SI_Del 63 utterances
in 20 minutes of interaction

Gesture production



FG_Ring 8 gestures
SI_Del 16 gestures
in 20 minutes of interaction

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)

	GG_Ring	FS_Ring
Mental Age (CPM)	04;03	06;00

Comparison 2 – Language Development

Types of different words produced in 15 minutes of interaction

	GG_Ring	FS_Ring
Type	46	140

Mean length of utterance

	GG_Ring	FS_Ring
MLU in words	2	2.71

Comparison 3 [LL_Del – BF_Del]

2 children with chromosome 14 deletions:

- **LL (07;08) M → Deletion**

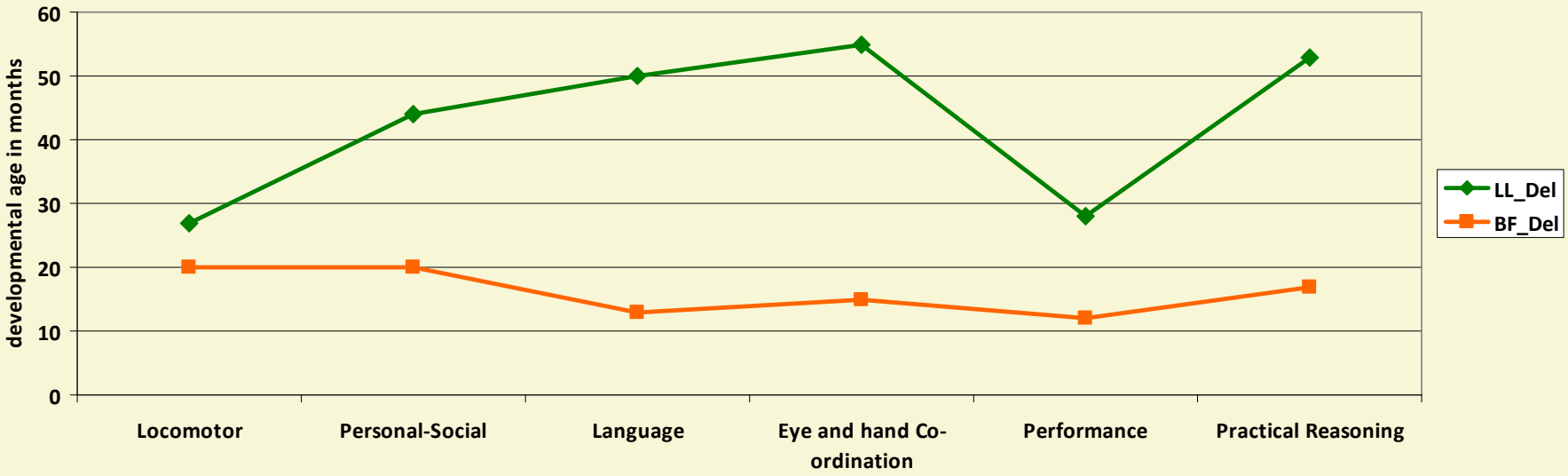
No autism/ No epilepsy/ minor cerebral morphological anomalies (thinning of the corpus callosum)

- **BF (07;08) F → Deletion**

No autism/ No epilepsy / several cerebral morphological anomalies (hypoplasia of the corpus callosum, opercular deficit)

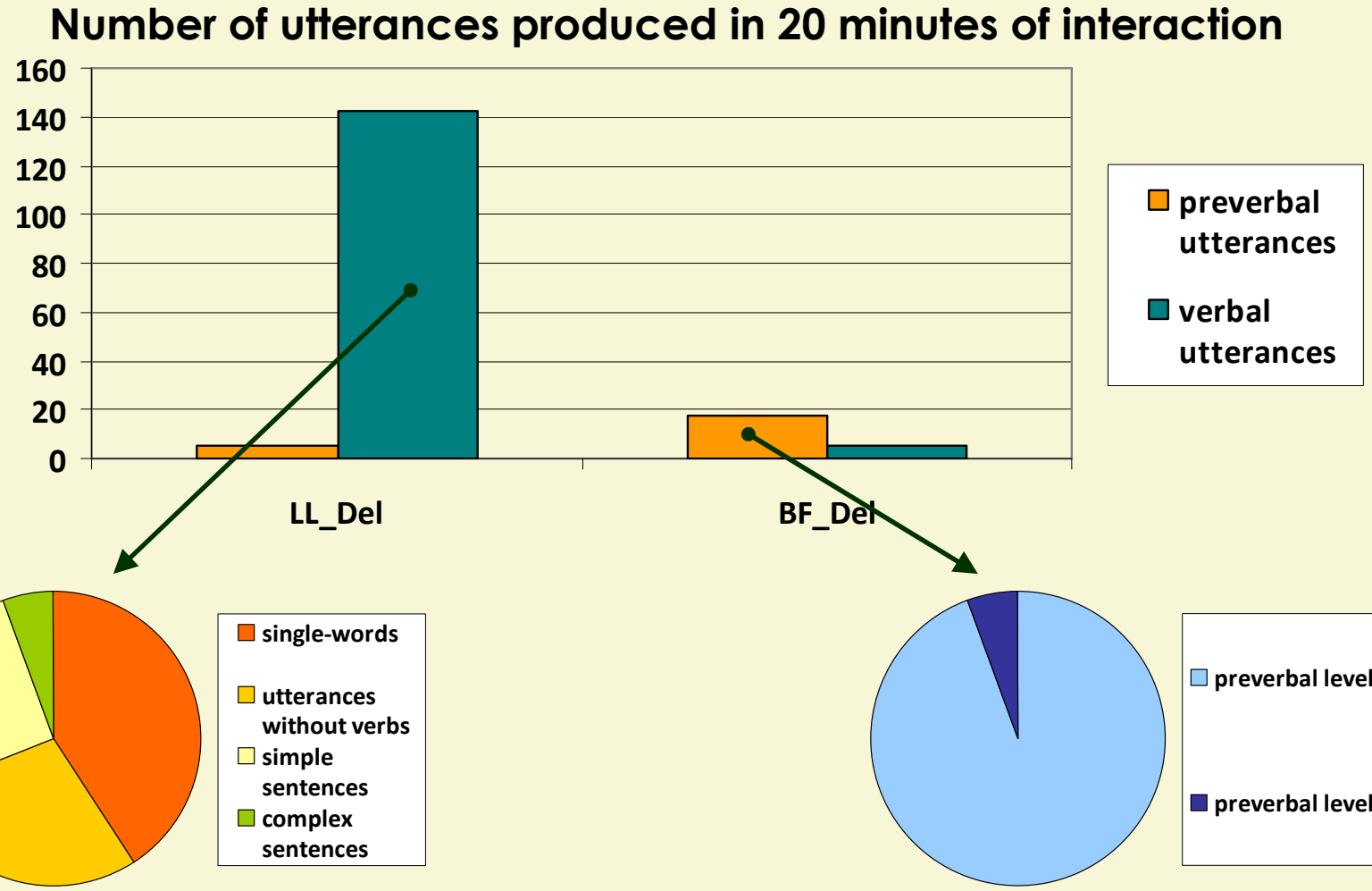
Comparison 3 – Psychomotor Development

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



Verbal utterance composition LL_Del

Preverbal utterance composition BF_Del

Conclusions

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**

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www.ring14.org

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IMPEGNO, SOSTEGNO, FUTURO.