

(laura.zampini1@unimib.it)

Linguistic and Psychomotor Development in Children with Chromosome 14 Deletions

Introduction

Chromosome 14 deletions are a specific type of rare genetic condition. In the literature, various aberrations involving chromosome 14 have been reported, such as the ring 14 syndrome (r14) and the linear 14q deletions.

Deletion of part of the genetic material on chromosome 14 (usually at the end of the long arm) causes various anomalies. Children with this genetic condition usually show developmental delays in addition to brain and neurological problems.

AIMS OF THE STUDY

- To describe and follow the language development of a group of children with linear 14q deletions during a one-year period.
- To examine to what extent individual differences in children's psychomotor or linguistic skills could be explained by the size or position of the deleted genetic material, as well as the presence of cerebral anomalies and autistic traits.

Method

PARTICIPANTS

Four children with chromosome 14 deletions:

	Gender	Age	Deleted area	Autistic traits
ш	М	7,5	10 Mb on 14q31.12q32.2	
BF	F	7,5	3 Mb on 14q32.2	
DMR*	F	6	18 Mb on 14q31.2q32.2	+
DMT*	F	6	18 Mb on 14q31.2q32.2	+

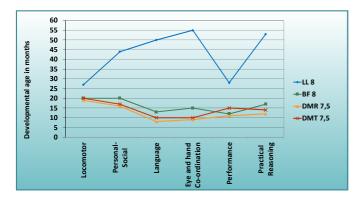
* DMR and DMT are monozygous twin sisters

PROCEDURE

Children's psychomotor development has been assessed by Griffiths Mental Development Scales-Extended Revised.

Their language development was assessed at baseline (T1) and then twice more in six-month intervals (T2 and T3) during mother-and-child free play sessions).

Results: Psychomotor development



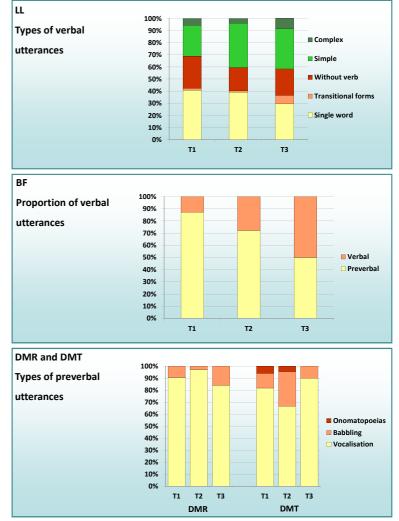
Results: Language development

The four children show very different levels of language development. LL's spontaneous production was almost exclusively verbal.

BF's production was characterized by a part of preverbal utterances and a part of single-word utterances.

DMR and DMT's vocal production was almost exclusively preverbal except for DMT's production of two words.

The longitudinal trend of children's language development is shown for each child in the figures below.



Conclusions

Data collected from psychomotor and linguistic assessments highlight a large individual variability.

Considering the children's genetic and clinical conditions, findings revealed that the size of the deleted area is not related to outcome.

However, the developmental trajectories of language development are deeply influenced by the presence of clinical conditions, such as autism spectrum disorders.