

# ***The fragile X syndrome : What about the deficit in the pragmatic component of language ?***

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# ***What is fragile X syndrome (FXS) ?***

- Most common inherited cause of mental retardation.
- 1 male per 2000 life birth / 1 female per 4000 life birth.
- Mutation on the X chromosome :
  - Break (or fragile site) at the bottom of the X chromosome.
  - Fragile X Mental Retardation 1 Gene (FRM1) discovered in 1991 (Verkerk & al.).
    - Repetitive trinucleotide sequence (CGG) found at the beginning of the FRM1 gene.
      - Non-FXS individuals : 5 to 50 CGG repeats → *normal*
      - FXS carriers : 53 to 200 CGG repeats → *premutation*
      - FXS individuals : more than 230 CGG repeats → *full mutation*

# *Phenotype of FXS males.*

*Wide spectrum of physical, behavioral, cognitive and language problems.*

## *Physical features*

- Long face
- Proeminent ears
- Soft and smooth skin
- Flat feet
- Craniofacial asymmetry
- Large testicles
- Hyperextensible finger joint
- Scoliosis

## *Behavioral features*

- Limited attention spans
- Hyperactivity
- Oversensitivity to tactile, auditory, olfactory and visual stimuli
- Avoid eye contact
- Autistic-like stereotypies (e.g., hand flapping, hand biting)

## *Cognitive features*

- MR in  $\pm$  85% of males with full mutation
- Mean IQ :
  - **41** for males with completely methylated full mutation
  - **60** for males with a mosaic pattern
  - **88** for males with an unmethylated or partially unmethylated full mutation

## ***The case FXS females.***

- Female with FXS are usually less affected than males.
  - Female typically have the full mutation on only one of their two X chromosomes → the unaffected chromosome moderates the effects of the mutation.

### ***Cognitive and behavioral consequences***

± 50 to 70% of the females with the full mutation have IQs in the borderline or mentally retarded range.

Females with the full mutation but without mental retardation have learning problems including executive function problems → can lead to limited attention spans and impulsivity.

# ***Speech production.***

- **Articulation :**
  - Omission, distortion and substitution of consonants and vowels in the conversational speech.
  - Errors reflecting the simplification processes observed in normally developing children.
- **Articulation rate :**
  - Variability in speaking rate → unpredictable shifts from rapid to slower rates.
- **Dysfluency :**
  - Breaks in the speech flow (including repetitions), inappropriated stops, interjections
- **Dysprosody :**
  - Litany.

# ***Lexical development.***

- Below chronological age expectations on both receptive and expressive measures of vocabulary.
- Some questions remain unsolved :
  - Are receptive and expressive vocabularies impaired to the same degree in affected males ?
  - What about the lexical development of FXS females ?
  - What about the strategies used in new words learning ?
  - What about the semantic categories and the lexico-grammatical categories acquisition ?

# ***Morphosyntactic development.***

- Below chronological age expectations on both receptive and expressive measures of morphosyntax.
  - Receptive morphosyntax is mental-age appropriate
  - Expressive morphosyntax : results are less clear
    - Paul & al. (1984) → delays in the morphosyntax of the conversational language relative to the nonverbal mental age.
    - Madison & al. (1986) →  $MLU \geq$  mental-age expectations.
    - Ferrier & al. (1991), Paul & al. (1987) → differences in expressive morphosyntax between FXS males and age- and cognitive-ability matched groups of males from several other diagnostic groups.
- ➔ Differences in the results may be attributable to variations in participants characteristics and small sample size.

# ***Communication and pragmatic development.***

- Below chronological age expectations for all domains of the VABS (Vineland Adaptive Behavior Scales) → scores closer to MA than to CA.
- Problems become more severe in adolescence
  - Scores on *Communication* and *Socialization* < those on *Daily Living Skills*.
  - The cause of this change is still unknown :
    - Because of the increase of unfamiliar people and setting in their environment ?
    - Because unfamiliar social situations become to stressfull ?
- Performances on communication tasks < those of developmental level matched mental retarded individuals (especially, autism and Down syndrome).



# Communication and pragmatic development.

*Perseveration → excessive self-repetitions of words, phrases, sentences or topics.*

## *FXS males without autism*

- Produce more self-repetitions than non-FXS males with autism
- *perseveration may be unique to FXS.*

## *FXS males without autism*

- Do not engage in echolalia (repetition of linguistic contribution of other people)
- *perseveration ≠ general tendency to repeat any previous behavior.*

# Causes of perseverations

## Four hypothesis

### *Deficient expressive morphosyntax*

- Strategy for participating in conversation when a failure to master morphosyntax make meaningful contribution impossible.
- Not supported by data

### *Word-retrieval deficit*

- Strategy emerging from the need to talk in the face of an inability to find the words needed to express a particular meaning.
- Supported by some data

### *Hyperarousal*

- Consequence of the arousal induced by various classes of stimuli especially those including interpersonal component
- Supported by some data

### *Executive function deficits*

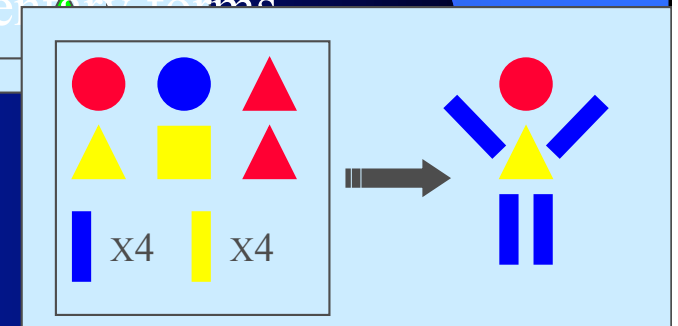
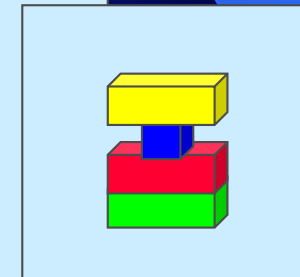
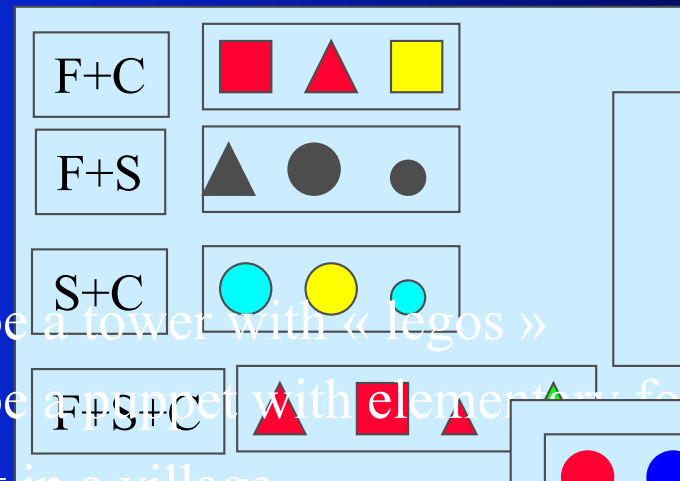
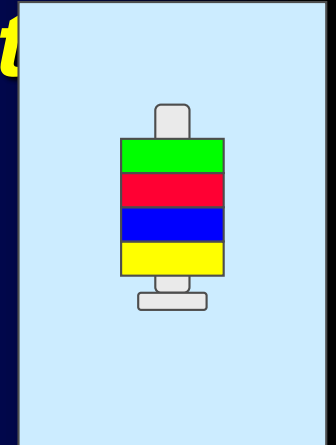
- Suspected in FXS males but difficult to measure.
- EFD reflect frontal lobe dysfunction and inhibition deficits
- Supported by neuroimaging data.

## ***Limitation of the recent researches on communication and pragmatic in FXS.***

- Few researches on communication and pragmatic development in FXS females.
- Assessment of FXS males almost exclusively :
  - Within the conversation context,
  - With a limited range of partners,
- No serious description of the ability of FXS to fulfill the requirements of the listener's role.
- Few studies on the emergence of the communicative problems of FXS individuals over the course of development.

# Our study : Referential communication in young FXS males.

- Sample : 4 FXS males aged 10;6 to 12;7 years-old
- Tasks :
  - Preliminary task : build / describe a tower with colored pearls
  - Task 1 : find / describe a particular combination
    - Form + color
    - Form + size
    - Size + color
    - Form + size + color
  - Task 2 : build / describe a tower with « legos »
  - Task 3 : build / describe a puppet with elementary forms
  - Task 4 : place a puppet in a village
  - Task 5 : find / describe a picture



# Subjects.

	<i>Chronological age (AC) in years</i>	<i>Lexical age (Peabody Picture Vocabulary Test)</i>
<b><i>Subject 1</i></b>	12;7	6;6
→ <i>Interlocutor</i>	10;4	6;6
→ <i>Matched typical child</i>	6;0	6;6
<b><i>Subject 2</i></b>	11;10	5;10
→ <i>Interlocutor</i>	11;5	5;10
→ <i>Matched typical child</i>	5;5	5;10
<b><i>Subject 3</i></b>	11;1	4;2
→ <i>Interlocutor</i>	14;1	4;2
→ <i>Matched typical child</i>	4;5	4;2
<b><i>Subject 4</i></b>	10;6	6;0
→ <i>Interlocutor</i>	10;10	6;0
→ <i>Matched typical child</i>	6;2	6;0

## *Situations.*

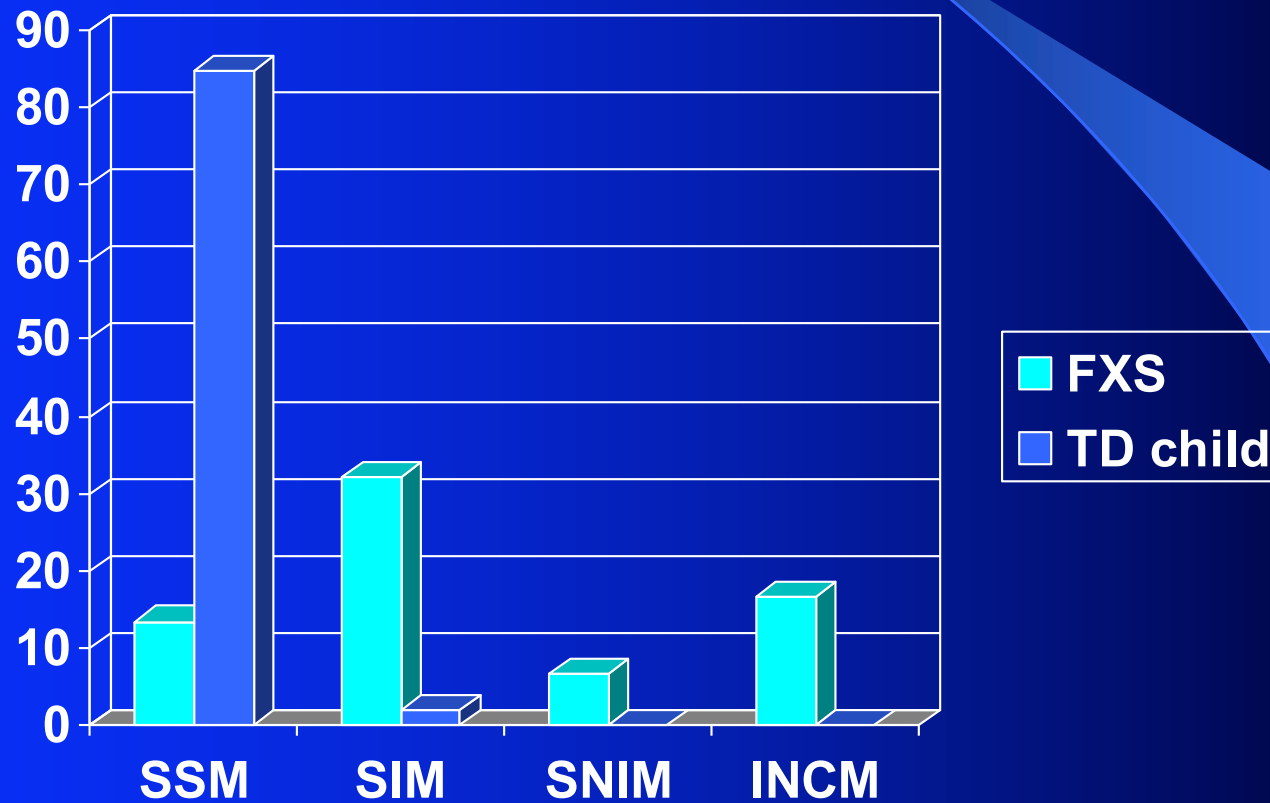
	<i>Speaker</i>	<i>Listener</i>
<i>Situation 1</i>	FXS child Typical child 1	OE child Typical child 2
<i>Situation 2</i>	OE child Typical child 2	FXS child Typical child 1
<i>Situation 3</i>	Adult (complete)	FXS child Typical child
<i>Situation 4</i>	Adult (incomplete)	FXS child Typical child

OE = other etiology

## ***FXS as speaker.***

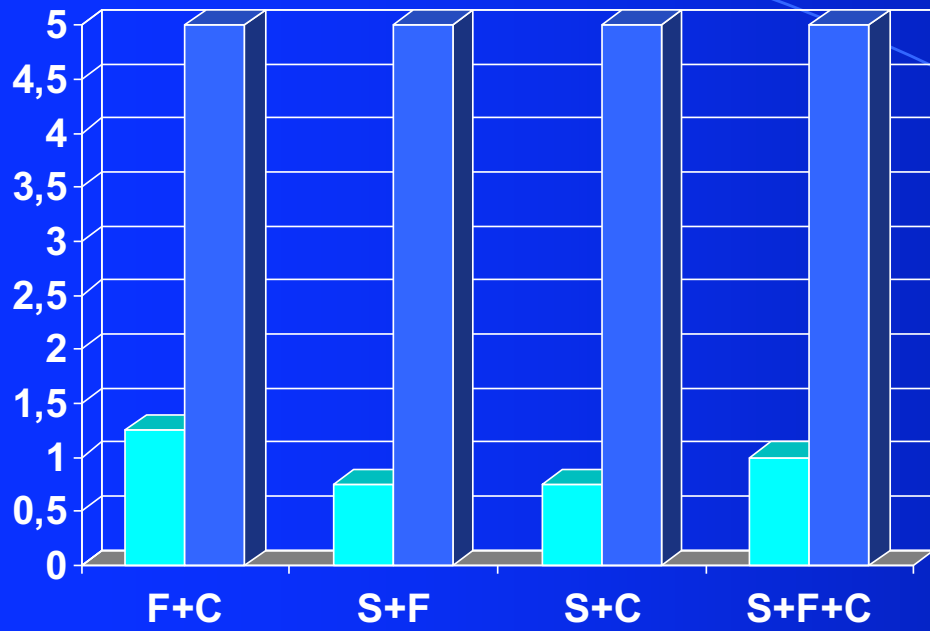
- Facing another MR child.
- Comparatively to typically developing children.
- In a first time, analysis of 4 kinds of messages :
  - *Spontaneous sufficient message* : containing all the information needed and generally leading to a correct response of the interlocutor.
  - *Spontaneous insufficient message* : containing not enough information; the listener must question the speaker to find the correct response.
  - *Spontaneous non-informative message* : no pertinent information is given concerning the item to describe (e.g., it's a puppet).
  - *Spontaneous incorrect message* : information given is incorrect (e.g. may concern a distractor).

# ***FXS poor communicators !***

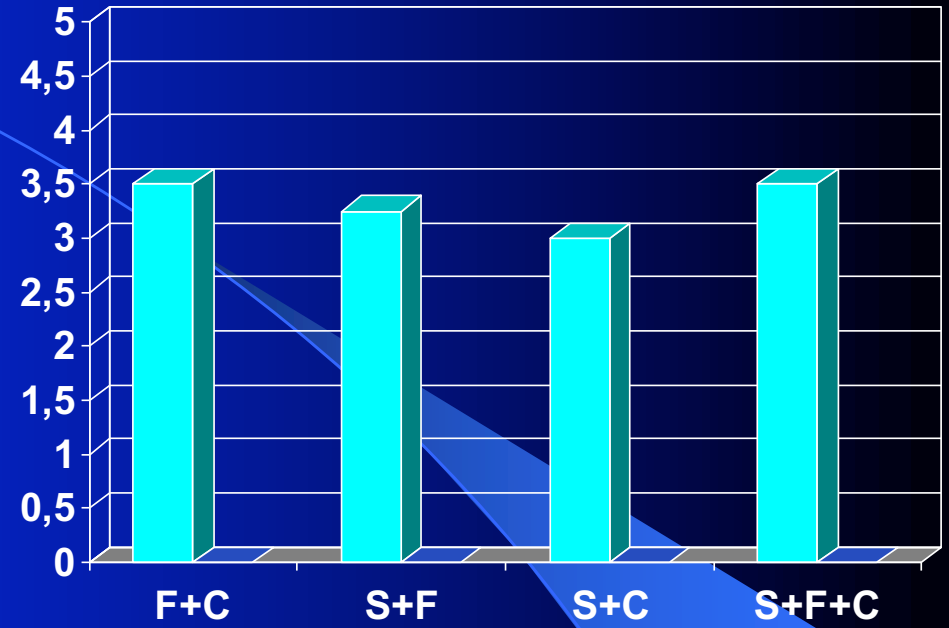




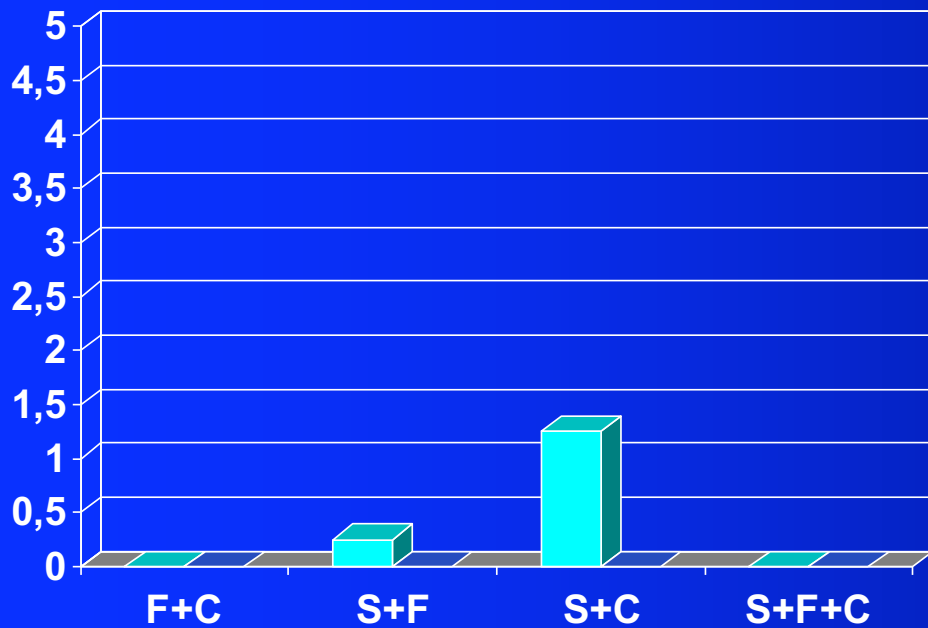
### Spontaneous sufficient messages



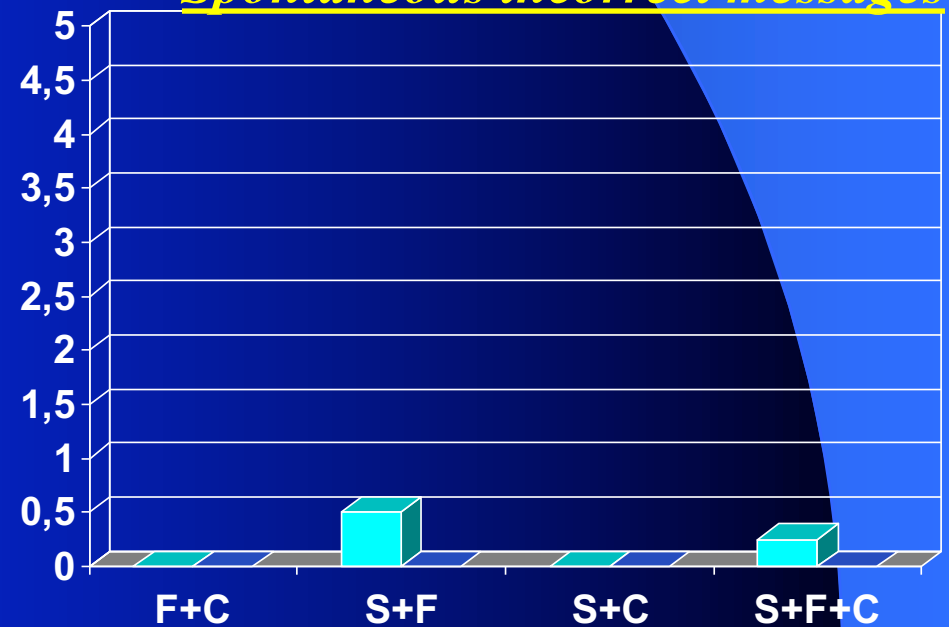
### Spontaneous insufficient messages



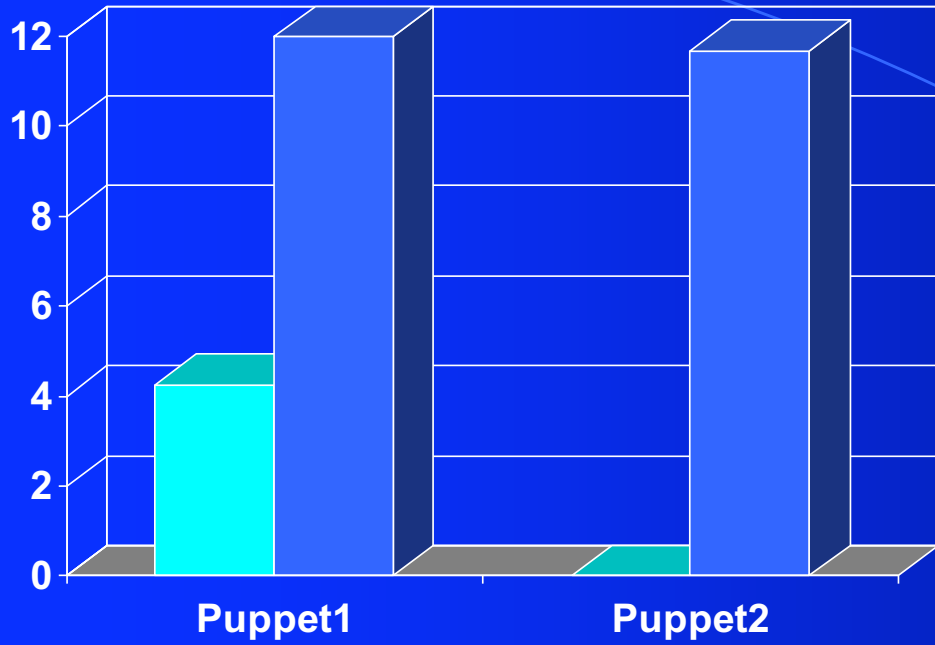
### Spontaneous non-informative messages



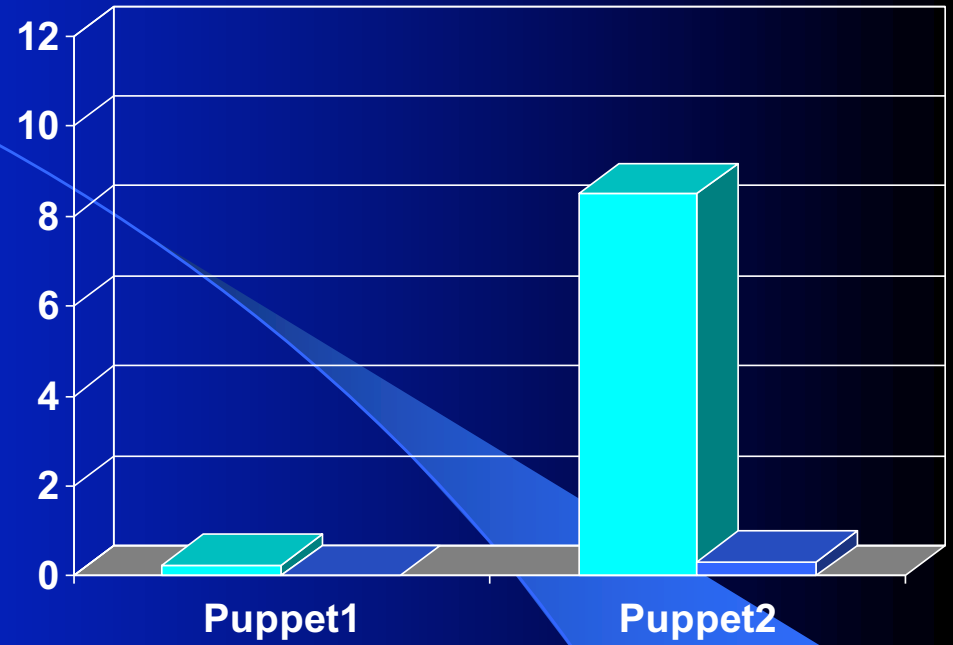
### Spontaneous incorrect messages



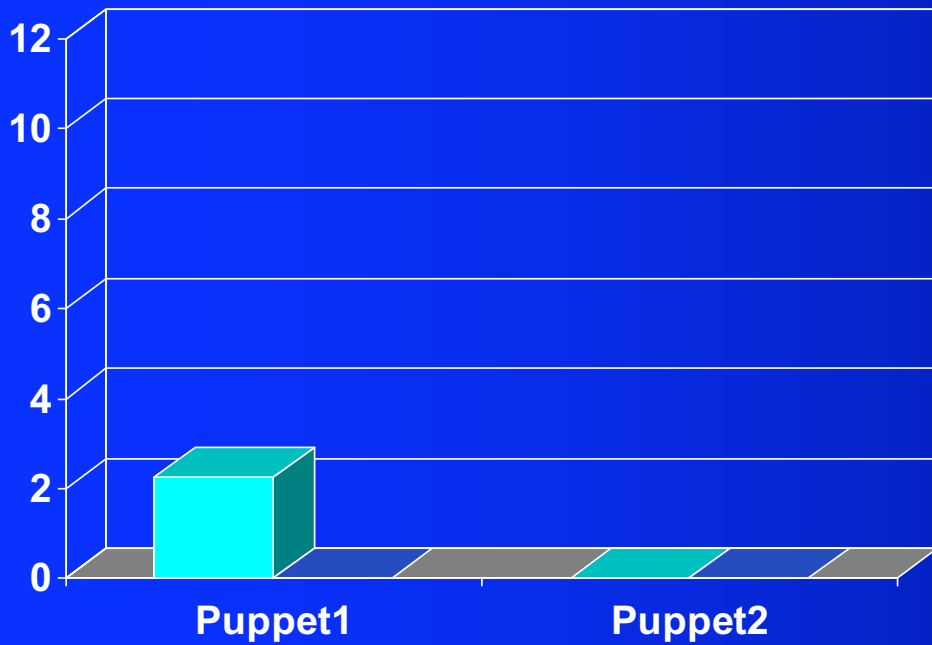
Spontaneous sufficient messages



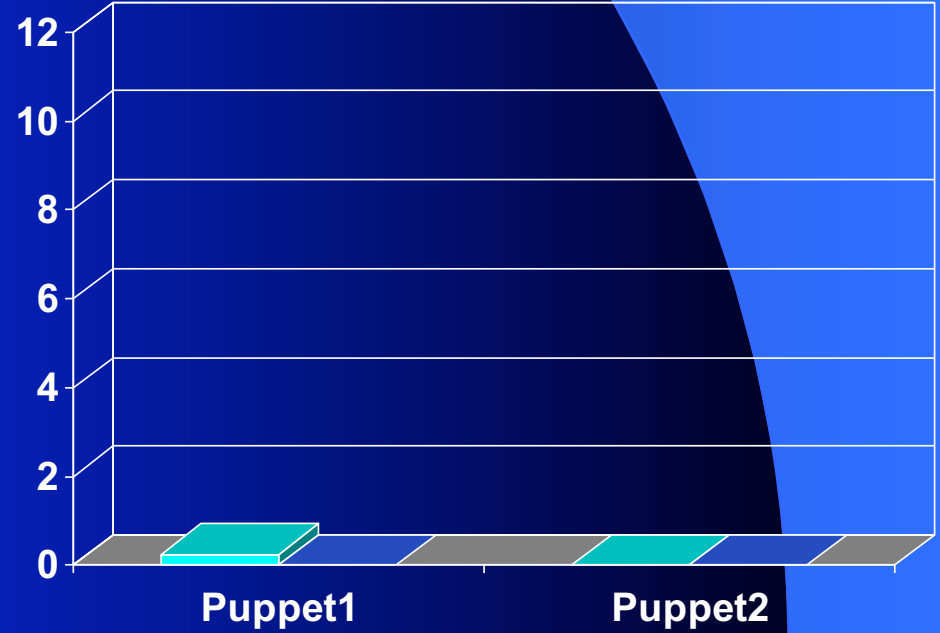
Spontaneous insufficient messages



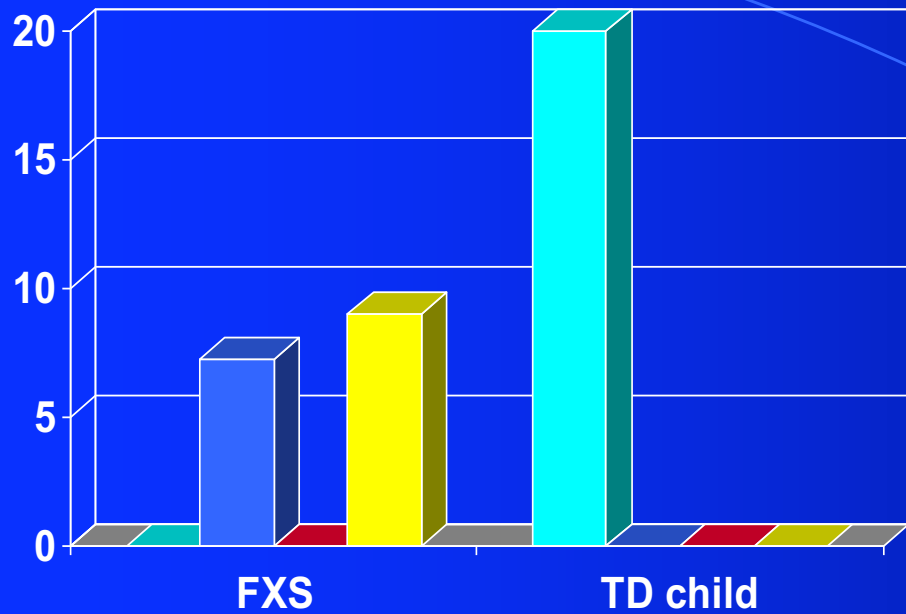
Spontaneous non-informative messages



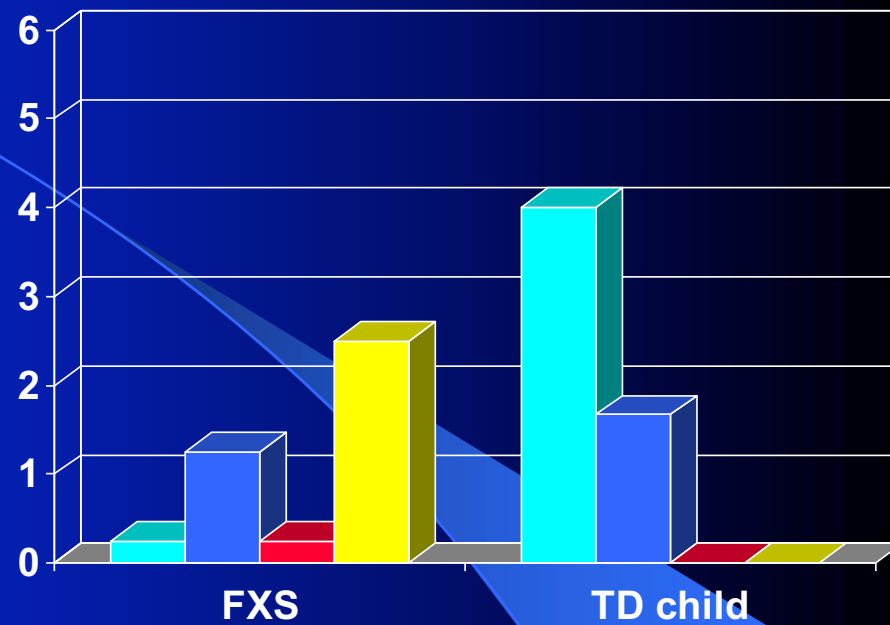
Spontaneous incorrect messages



### Legos

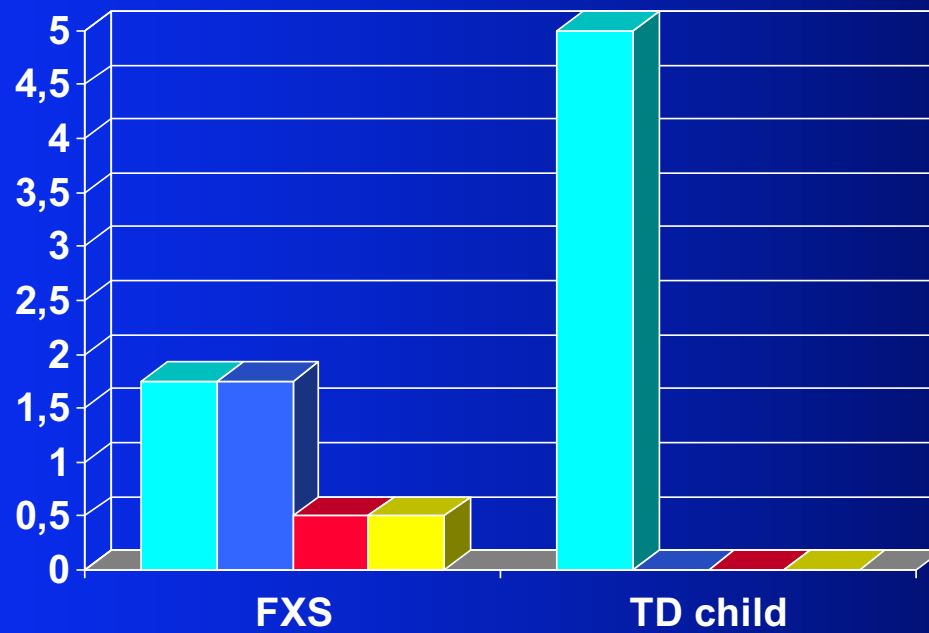


### Village

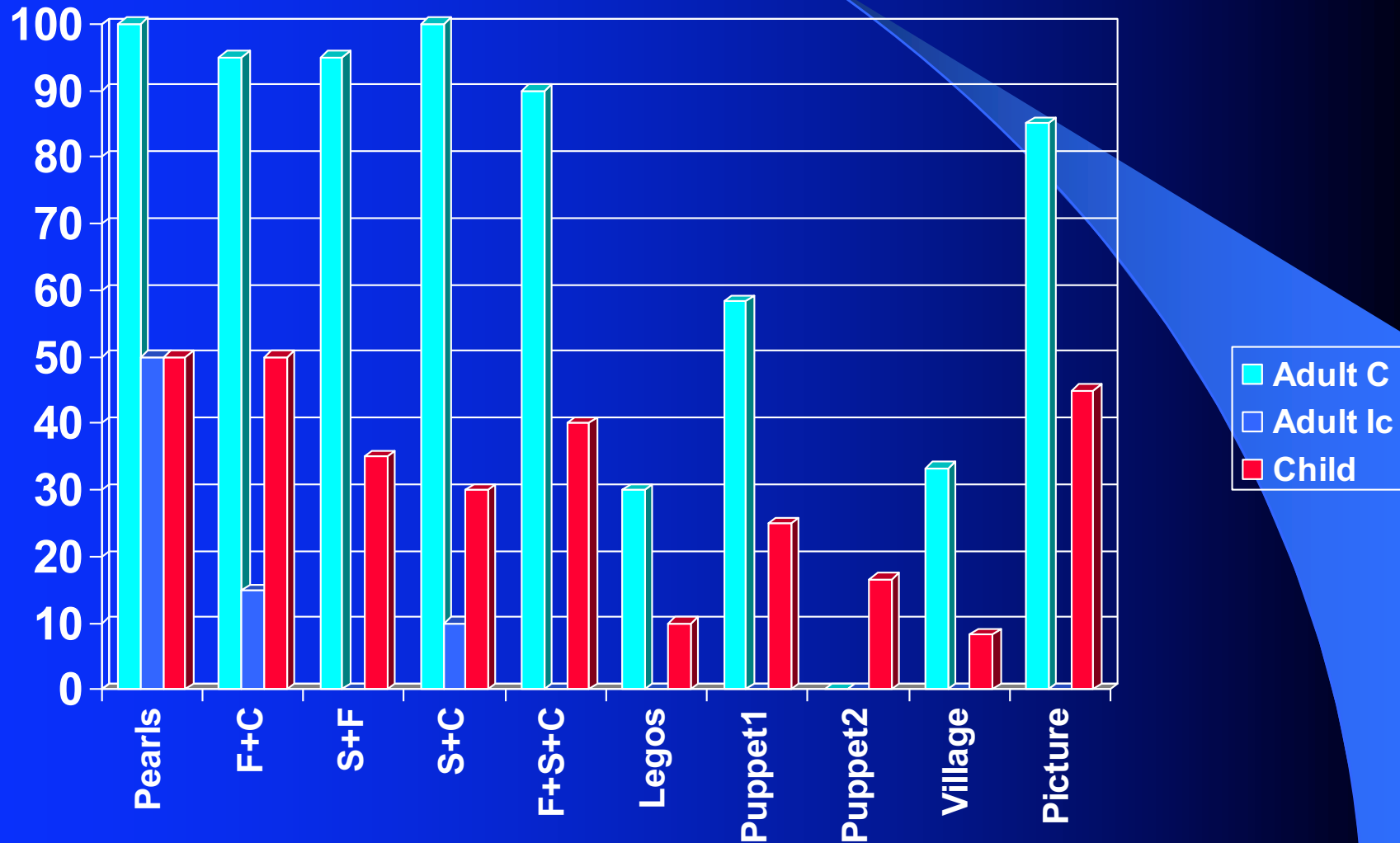


■ SSM ■ SIM ■ SNIM ■ SIncM

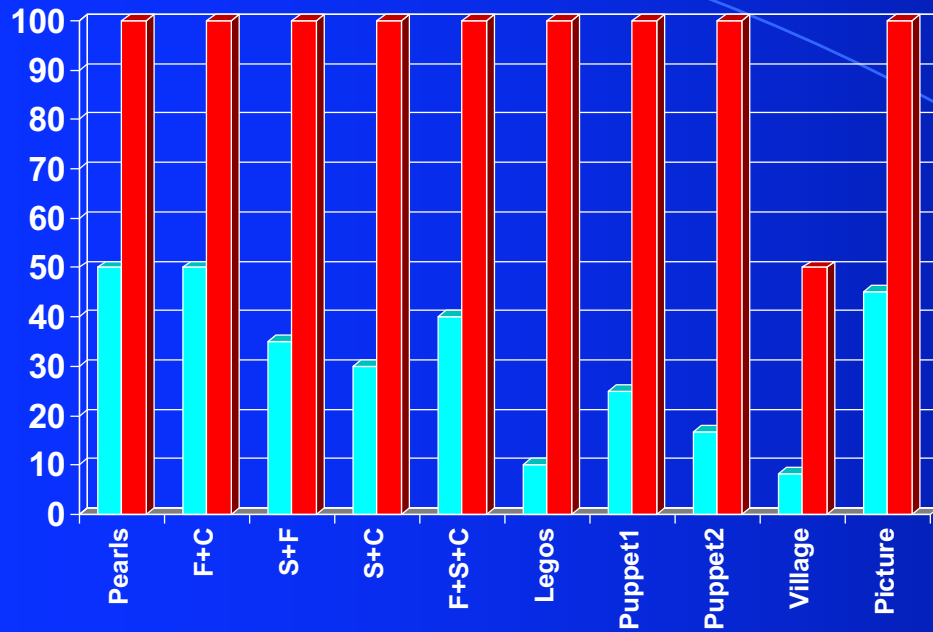
### Picture



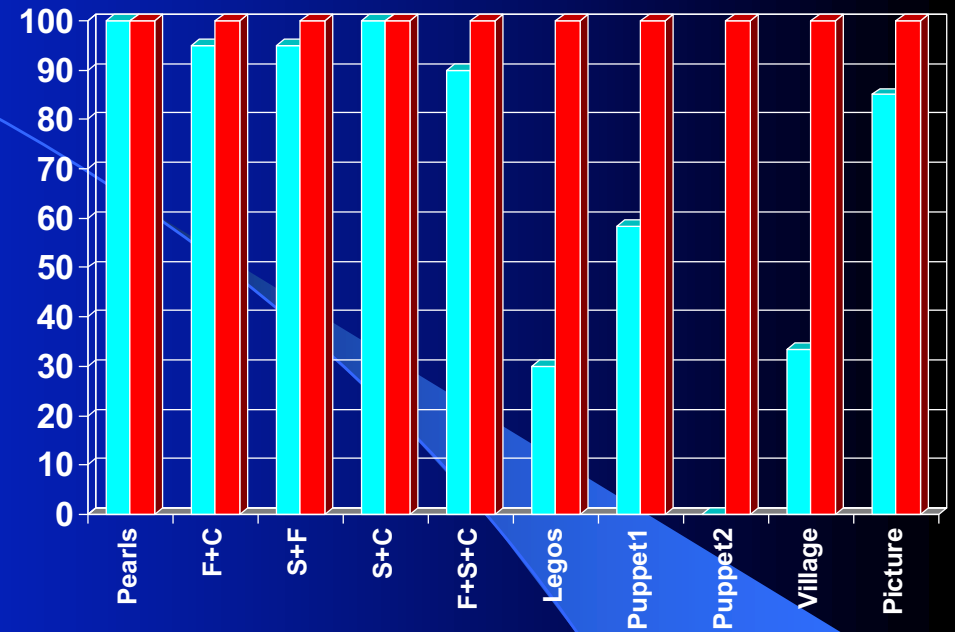
## ***FXS poor listeners ?***



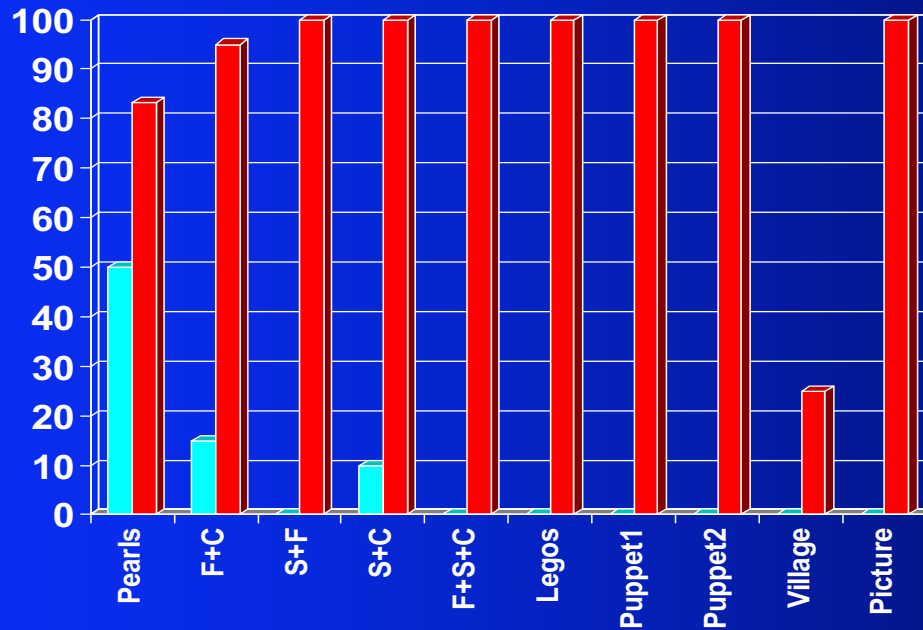
## Facing another child



## Facing a « complete adult »



## Facing an « incomplete adult »



# ***Preliminary conclusions.***

## *Speaker*

- FXS give more spontaneous insufficient messages than TDC.
- Messages are rarely totally inappropriate or incorrect.
- Difficulties with spatial information.
- It doesn't seem to be a morpho-syntactic deficit but rather :
  - Difficulty in finding the right word.
  - Difficulty in finding the pertinent features.

## *Listener*

- FXS perform as well as TDC with a « complete adult » except for the task containing spatial information or for building tasks.
- FXS are less performant with a child or an « incomplete adult »
- FXS engage more easily in a verbal interaction with a child than with an adult.

## *In progress.*

- Enlarged the actual sample.
- Using an « eye contact » condition
  - Do they engage in a tangential language ?
  - Do they present more perseverations ?
  - Do we observe a dramatic decrease of performance ?
- Comparisons with other etiological groups characterised by pragmatic disorders.