



# Recognition of Facial Expressions of Emotions and Theory of Mind in Children with Neurofibromatosis Type 1

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# Neurofibromatosis type 1 (NF1)

- Genetic neurocutaneous syndrome (*Hachon et al., 2011*)
  - Prevalence 1/3500
  - Physical feature = café-au-lait spots
- Prevalence (*Hyman, 2005*)
  - Distribution of total IQ = normal
  - Learning disabilities = 52 %
  - Attention deficit hyperactivity disorder (ADHD) = 38 %

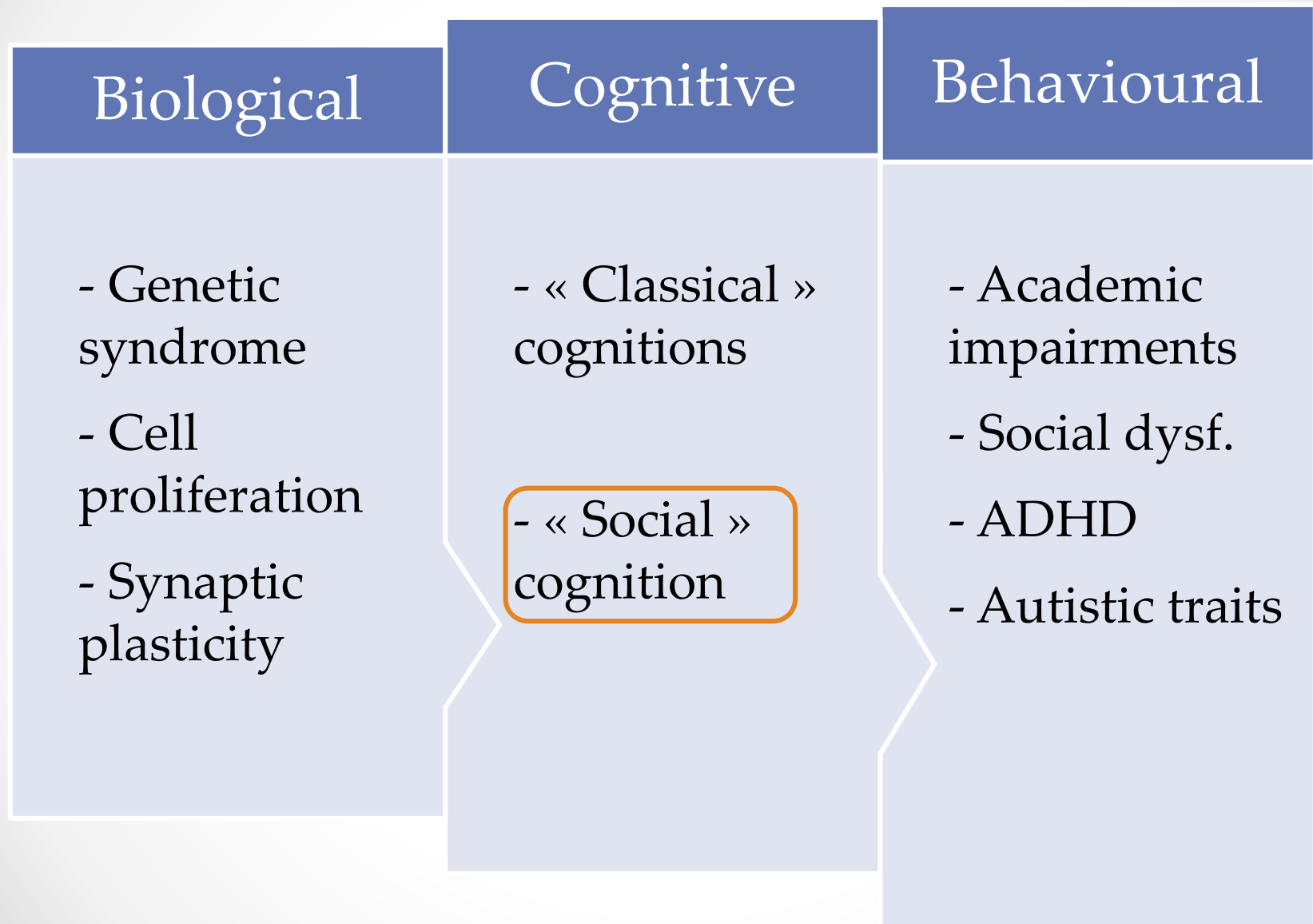


*Haymanj (2007) wikimédia*

# Neurofibromatosis type 1 (NF1)

- Social dysfunction (*Noll, 2007*)
  - Isolation and poor social skills
  - Reduced social outcome
  - Prevalence of autistic traits > 30 % (*Plasschaert, 2015*)
- Quality of life (*Gilboa et al., 2010*)

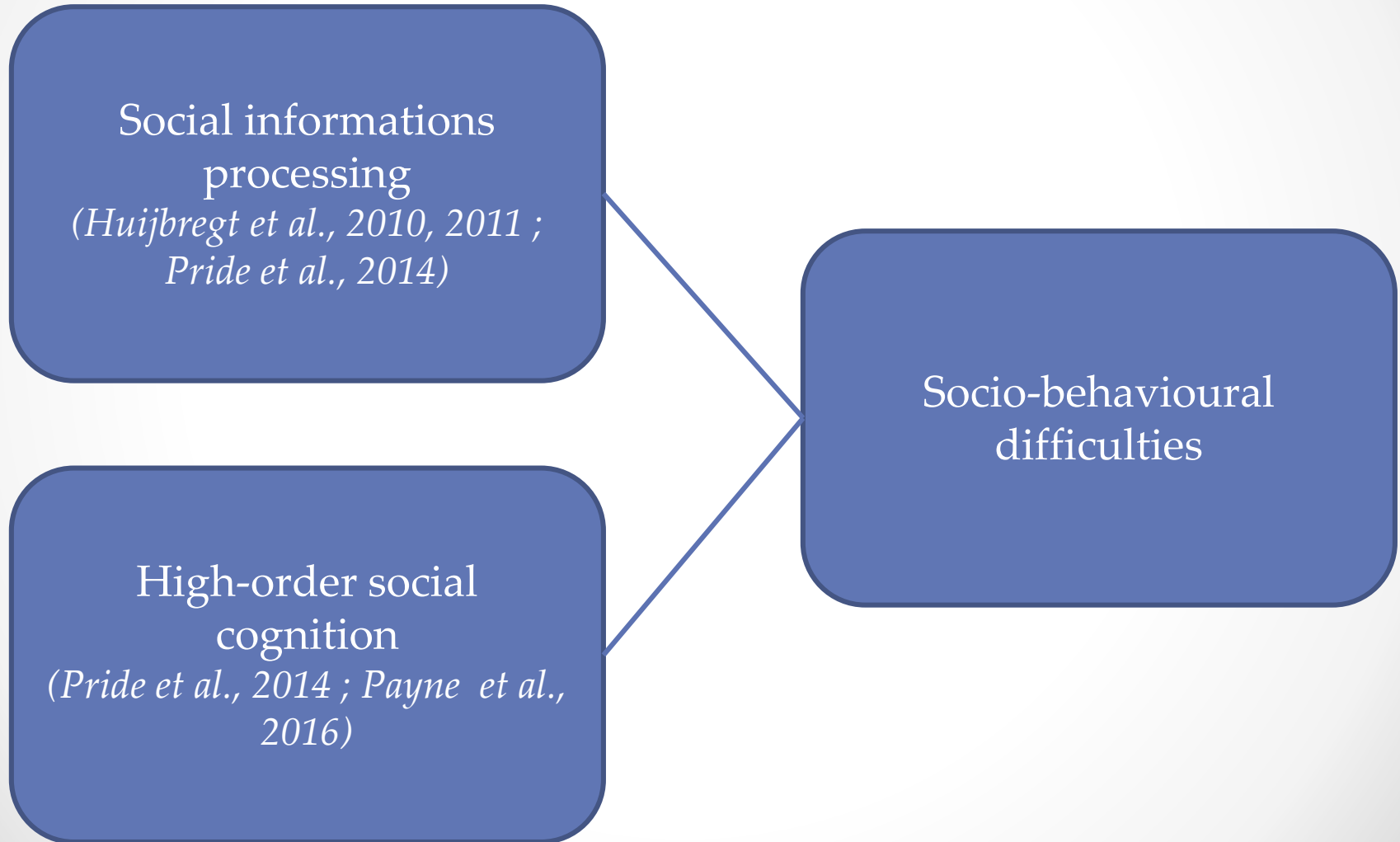
# Neurofibromatosis type 1 (NF1)



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## Cognitive level

## Behavioural level



# Issue

- **Recognition of emotions** of fear and anger seems to be difficult for subjects with NF1, but
  - Only 2 studies,
  - We don't know if difficulties start before 8 years old
  - > Needs to take into account children between 4 to 12 years old
- **High-order social cognition**
  - Only 1 in 2 studies investigated Theory of Mind (ToM)
  - Non verbal tasks, few items on false belief
  - > Needs to take into account
    - reasoning on false belief (cognitive ToM)
    - inference on emotional states based on false belief (affective ToM)

# Hypotheses

- **1 / recognition of facial expressions of emotions (RFE)**
  - Mean score NF1 < Control, especially for fear and anger
  
- **2 / ToM**
  - Mean score NF1 < Control, for ToM
  - NF1 : positive correlations ToM & Inhibition (INH) and Working Memory

# Methodology

## Participants aged 4 to 12

NF1 (N = 28) <sup>1</sup>		Control (N = 28)		
Sex ratio (H : F)	15 : 13	15 : 13		
T test	Moy. (S.D)	Moy. (S.D)	<i>t</i>	<i>p</i>
Age in month	93.32 (29.37)	94.11 (29.30)	0.1	.92
Verbal abstraction (Sim. WISC)	27 (7.99)	30.39 (9.33)	1.46	.15

<sup>1</sup> Diagnostics made by a pediatric neurologist



# Methodology

## Social cognition tasks

- ToM task battery  
(*Hutchins et al, 2008*)
- Recognition of facial expressions of emotions (*adapt. Nepsy II*)



Voici Julien. Julien lit un livre.



Une fois qu'il a fini de lire, Julien pose son livre sur la table.



Julien va chercher quelque chose à manger dans la cuisine.



Regarde, Sonia arrive et prend le livre qui est sur la table pour le mettre dans un tiroir.



Ensuite, Sonia s'en va.



Regarde, Julien revient pour lire son livre à nouveau.

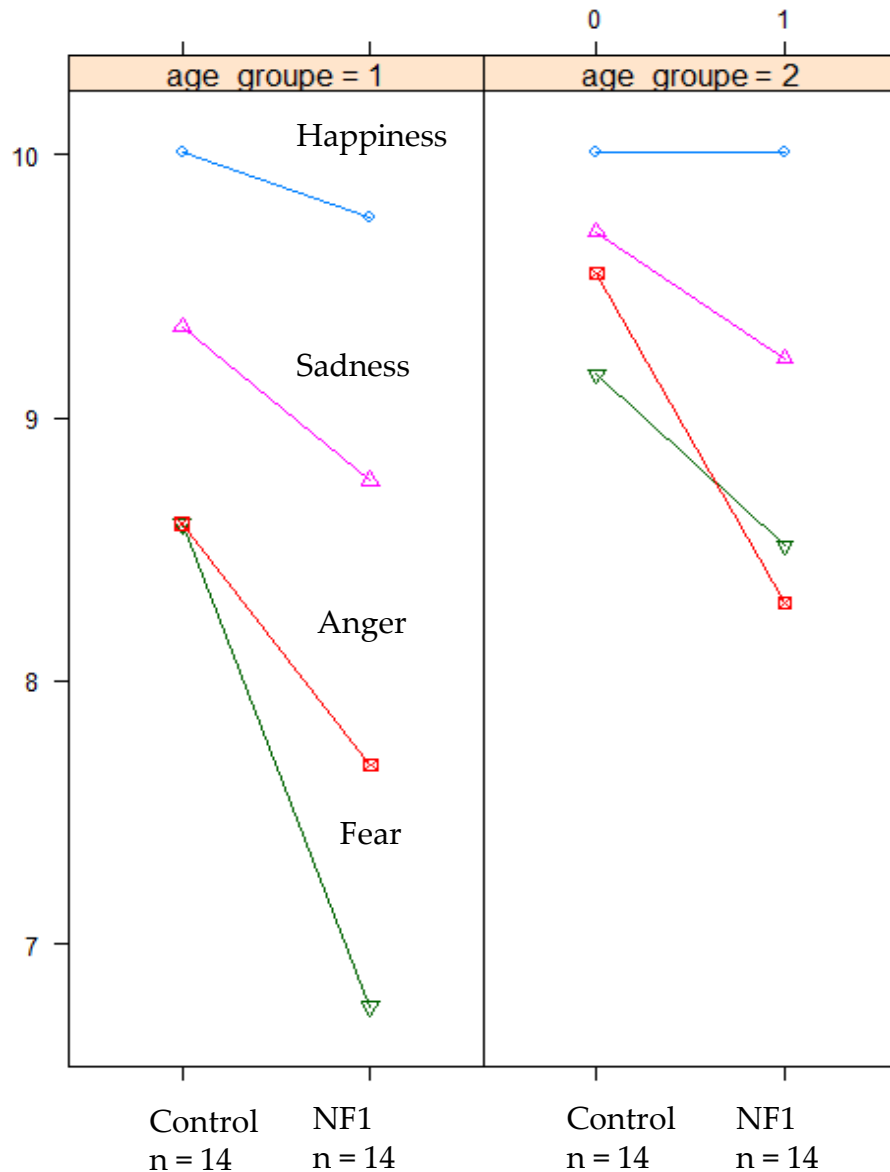
Where will Julien search his book?

# Methodology

## Control tasks

- Language (*Voc et Sim / WISC IV*)
- Working memory (*backward digit span / WISC IV; backward Corsi block tapping task*)
- Cognitive inhibition (*Nepsy II*)

# Results : emotion hypothesis

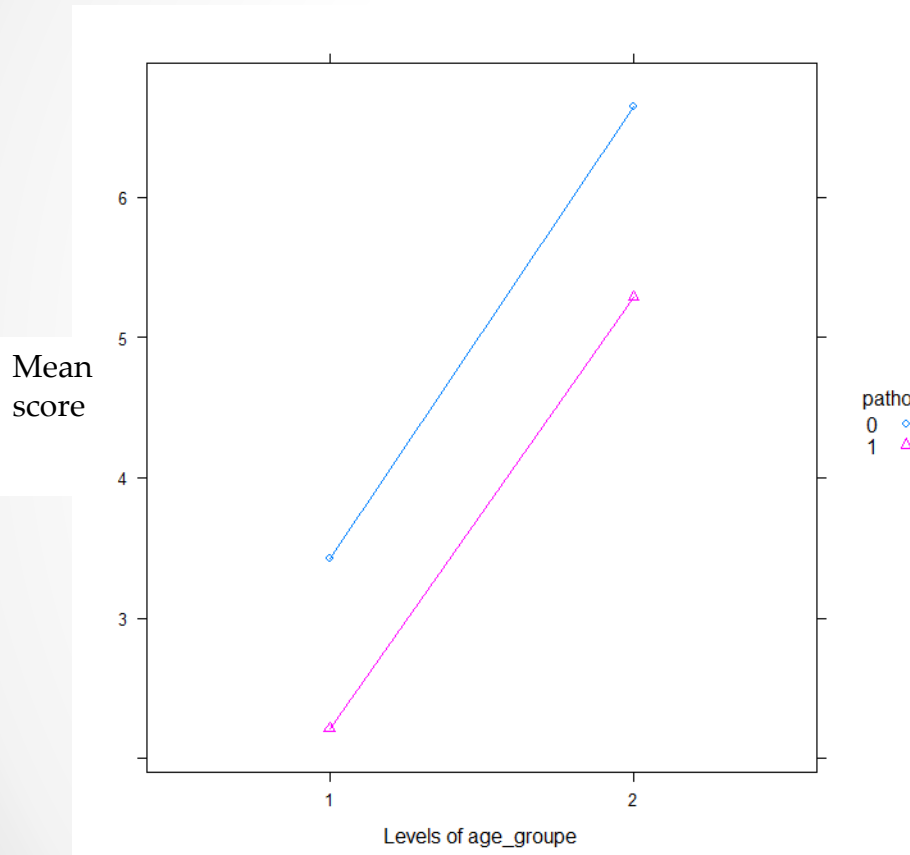


## ANOVA

	F	CME	ges	p
Patho	11.7	2.02	.09	<.01**
Age	14.5	2.02	.11	<.01**
Emot°	3.17	0.83	.03	<.05*

Age group 1 < 7 ½ years old  
Age group 2 > 7 ½ years old

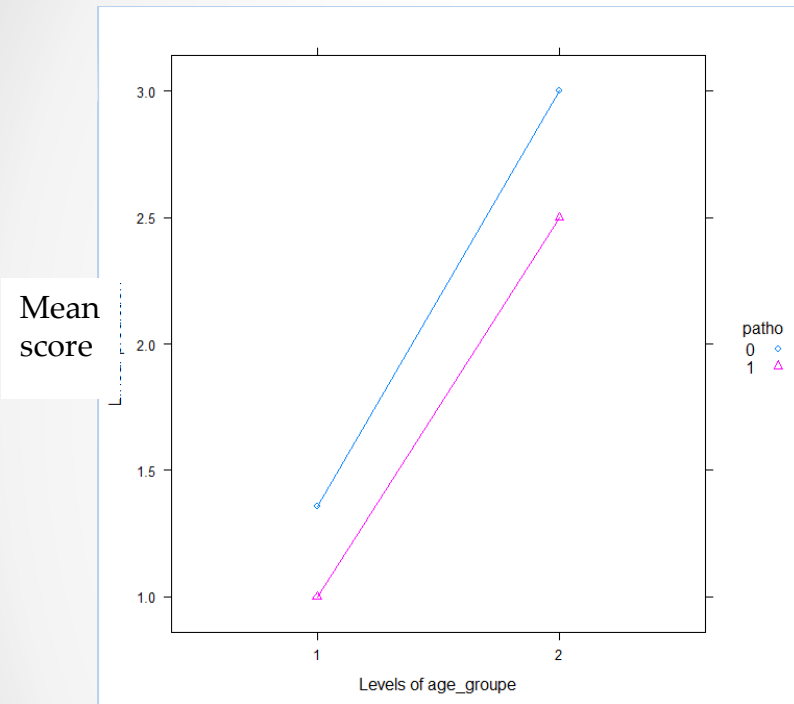
# Results : ToM hypothesis



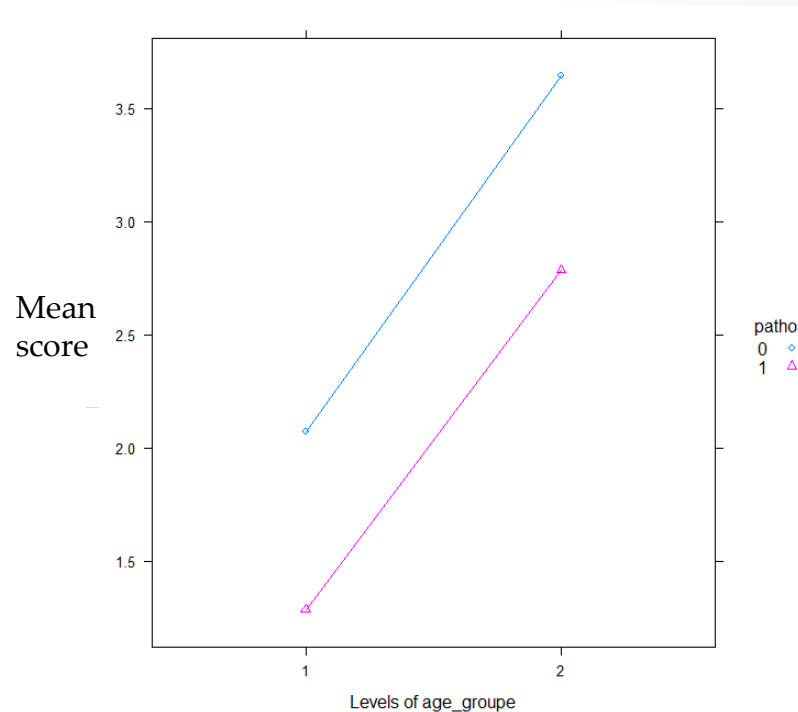
	F	CME	ges	<i>p</i>
Patho	10.7	2.15	.09	**
Age	64.3	2.15	.55	***

# Results : ToM hypothesis

## Affective ToM



## Cognitive ToM



	F	CME	ges	<i>p</i>
Patho	2.99	0.86	.05	ns
Age	40.2	0.86	.44	***

	F	CME	ges	<i>p</i>
Patho	13.2	0.72	.20	***
Age	45.9	0.72	.47	***

# Results : ToM hypothesis

- Partial correlation (control for age)

	NF1 : Total ToM	Control : Total ToM
WM	$r = .30$ $r^2 = .09$ $p = .11$	$r = .37$ $r^2 = .13$ $p = .05^*$
INH	$r = .05$ $r^2 = .004$ $p = .10$	$r = .53$ $r^2 = .28$ $p = .004^{**}$

# Discussion

- **1 / Recognition of Facial Expressions of emotions (RFE)**
  - Mean score NF1 < Control, especially for fear & anger
- These outcomes are consistent with the literature
  - Difficulties from age 4, for recognition of negative emotions -> developmental delay ?
  - What consequences ?
  - What causes ?

# Discussion

- **2a / ToM**

- NF1 < Control / ToM

=> this outcome corresponds well with the literature

- Mainly / cognitive false belief -> developmental delay?
- Similarities with Pride et al. /sarcasms
- What consequences ?
- What causes ?



# Discussion

- **2a / ToM**

- NF1 < Control / ToM

⇒ this outcome are consistent with the literature

- Mainly / cognitive false belief -> developmental delay?
- Similarities with Pride et al. /sarcasms
- What consequences ?
- What causes ?

- **2b / ToM**

- NF1 : no correlations ToM & Inhibition (INH) and Working Memory

- ≠ / literature regarding healthy child
- heterogeneity of NF1 group

# Limitations and requirements

- **1<sup>st</sup> statistical analysis**
  - Statistical procedures using mean of groups don't bring the heterogeneity of subjects in the NF1 group
  - ⇒ Data will be refined, probably with the LME procedure
- **recognition emotion task**
  - Non ecological material (frozen pictures) + uncontrolled response time + uncontrolled face expressiveness thresholds
  - ⇒ A tool should be used such as the TASIT / video
  - ⇒ Measuring response time and eye-tracking ?
- **ToM**
  - To develop scenarios / emotions of fear and anger to investigate affective ToM ?

# Conclusion

- Risk of deficit of RFE / fear, anger and sadness for NF1
- Risk of deficit of ToM for NF1
- First result that brought many questions -> further investigations are needed
- Relevance of an early screening of social cognition and, if required, of cognitive remediation

Thanks for your attention