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

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With the collaboration of MD P. Sabouraud
Neurofibromatosis type 1 (NF1)

• Genetic neurocutaneous syndrome *(Hachon et al., 2011)*
  o Prevalence 1/3500
  o Physical feature = café-au-lait spots

• Prevalence *(Hyman, 2005)*
  o Distribution of total IQ = normal
  o Learning disabilities = 52%
  o Attention deficit hyperactivity disorder (ADHD) = 38%
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)

<table>
<thead>
<tr>
<th>Biological</th>
<th>Cognitive</th>
<th>Behavioural</th>
</tr>
</thead>
<tbody>
<tr>
<td>- Genetic syndrome</td>
<td>- « Classical » cognitions</td>
<td>- Academic impairments</td>
</tr>
<tr>
<td>- Cell proliferation</td>
<td>- « Social » cognition</td>
<td>- Social dysf.</td>
</tr>
<tr>
<td>- Synaptic plasticity</td>
<td></td>
<td>- ADHD</td>
</tr>
<tr>
<td></td>
<td></td>
<td>- Autistic traits</td>
</tr>
</tbody>
</table>
Neurofibromatosis type 1 (NF1)

Cognitive level

- Social informations processing
  *(Huijbregt et al., 2010, 2011; Pride et al., 2014)*

- High-order social cognition
  *(Pride et al., 2014; Payne et al., 2016)*

Behavioural level

- Socio-behavioural difficulties
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)
  o Mean score NF1 < Control, especially for fear and anger

• 2 / ToM
  o Mean score NF1 < Control, for ToM
  o NF1 : positive correlations ToM & Inhibition (INH) and Working Memory
# Methodology

**Participants aged 4 to 12**

<table>
<thead>
<tr>
<th></th>
<th>NF1 (N = 28) ¹</th>
<th>Control (N = 28)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sex ratio (H : F)</td>
<td>15 : 13</td>
<td>15 : 13</td>
</tr>
<tr>
<td>T test</td>
<td>Moy. (S.D)</td>
<td>Moy. (S.D)</td>
</tr>
<tr>
<td>Age in month</td>
<td>93.32 (29.37)</td>
<td>94.11 (29.30)</td>
</tr>
<tr>
<td>Verbal abstraction</td>
<td>27 (7.99)</td>
<td>30.39 (9.33)</td>
</tr>
</tbody>
</table>

¹ 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*)
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**

<table>
<thead>
<tr>
<th></th>
<th>F</th>
<th>CME</th>
<th>ges</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patho</td>
<td>11.7</td>
<td>2.02</td>
<td>.09</td>
<td>&lt;.01**</td>
</tr>
<tr>
<td>Age</td>
<td>14.5</td>
<td>2.02</td>
<td>.11</td>
<td>&lt;.01**</td>
</tr>
<tr>
<td>Emot°</td>
<td>3.17</td>
<td>0.83</td>
<td>.03</td>
<td>&lt;.05*</td>
</tr>
</tbody>
</table>

Age group 1 < 7 ½ years old
Age group 2 > 7 ½ years old
Results: ToM hypothesis

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<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patho</td>
<td>10.7</td>
<td>2.15</td>
<td>.09</td>
<td>**</td>
</tr>
<tr>
<td>Age</td>
<td>64.3</td>
<td>2.15</td>
<td>.55</td>
<td>***</td>
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</table>
Results: ToM hypothesis

Affective ToM

<table>
<thead>
<tr>
<th></th>
<th>F</th>
<th>CME</th>
<th>ges</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patho</td>
<td>2.99</td>
<td>0.86</td>
<td>0.05</td>
<td>ns</td>
</tr>
<tr>
<td>Age</td>
<td>40.2</td>
<td>0.86</td>
<td>0.44</td>
<td>***</td>
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</tbody>
</table>

Cognitive ToM

<table>
<thead>
<tr>
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<th>F</th>
<th>CME</th>
<th>ges</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Patho</td>
<td>13.2</td>
<td>0.72</td>
<td>0.20</td>
<td>***</td>
</tr>
<tr>
<td>Age</td>
<td>45.9</td>
<td>0.72</td>
<td>0.47</td>
<td>***</td>
</tr>
</tbody>
</table>
Results: ToM hypothesis

- Partial correlation (control for age)

<table>
<thead>
<tr>
<th></th>
<th>NF1: Total ToM</th>
<th>Control: Total ToM</th>
</tr>
</thead>
<tbody>
<tr>
<td>WM</td>
<td>r = .30</td>
<td>r = .37</td>
</tr>
<tr>
<td></td>
<td>r² = .09</td>
<td>r² = .13</td>
</tr>
<tr>
<td></td>
<td>p = .11</td>
<td>p = .05 *</td>
</tr>
<tr>
<td>INH</td>
<td>r = .05</td>
<td>r = .53</td>
</tr>
<tr>
<td></td>
<td>r² = .004</td>
<td>r² = .28</td>
</tr>
<tr>
<td></td>
<td>p = .10</td>
<td>p = .004 **</td>
</tr>
</tbody>
</table>
Discussion

• 1 / Recognition of Facial Expressions of emotions (RFE)
  o 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
  o 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
  o NF1 : no correlations ToM & Inhibition (INH) and Working Memory
  ➢ ≠ / literature regarding healthy child
  ➢ heterogeneity of NF1 group
Limitations and requirements

• **1\text{st} 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