AUTISM SPECTRUM DISORDER AND VISUAL IMPAIRMENT

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Context

Regional professionnel network in Rhône Alpes :
ASD & Sensory Impairment

National network : France ASD-VI since 2017

RESEARCH: High prevalence of ASD in Blind and Severely Visually impaired children. Several studies estimate occurrence up to 30-50%

CLINICAL OBSERVATIONS: diagnosis of ASD complex to make – overlapping of clinical signs + lack of tests adapted to these children.

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Plan

- Characteristics specific to the development of visually impaired children
- Links to autism
- Developmental trajectories in visual impairment
- Symptom overlap and difficulty in reliably identifying ASD in VI children
- Presentation of a research project on very young VI infants
- Current questions and discussion of the issues

Early years development of children with congenital blindness or severe VI : Delays and atypical factors

Motor delays

Stéréotypic movements - “blindismes”

Characteristics of non verbal communication

Verbal language delays and characteristics

Atypical play behaviours

Delays or difficulties in social interactions

Challenges in the perception of Sensory information

Delays in motor development

Bullinger & Mellier (1988); Hatwell (2005); Fraiberg, (1977); Portalier (2001)

- Very little spontaneous exploration of the environnement in the first two years

- Delays in using the seated position independantly
- Difficuties changing physical positions (posture control)
- Delayed acquisition of walking, difficulties in coordinating walking skills (stiffness, hesitation…)
- Later establishment of object permanence
- HOWEVER : wide range observed, interindividual variability ++

Stereotypic movements- “blindismes”

Fazzi et al (1999) ; Smith et al, (1969) ; Tröster et al (1991) : McHugh & Lieberman (2003)

- Eye pressing (espcially in congenitally blind infants)
- Increased stereotypies when VI comorbid with learning disability
- Fazzi (1999) : at least one form of stereotypic movement in all severely VI individuals
- Rocking behaviours

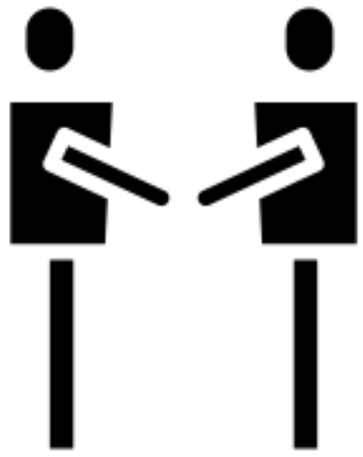
Functional sensory stimulation- adapting to the lack of visual input –

Some high risq situations (lack of stimulation, stressful environment…)

Atypical non-verbal communciation

Mellier & Jouen (1984) ; Loots et al (2003) ; Portalier (2001)

- Non-verbal communication with unusual signs (ex: becoming still or lowering the head to concentrate on auditory information)
- Original signals to attract others' attention
- Lack of pointing and hand gestures
- Less facial expressions
- Difficulties in interpreting others' emotional expressions



“Delays” in langage and atypical verbal communciation

Galiano et al (2019); Fraiberg (1977) ; Fazzi et al (2007) ; Priesler (1993) ; Mellier & Deleau (1991); Perez-Pereira & Castro (1997);

- Less frequent social initiatives, less frequent vocalisations

- Pragmatic language differences :
  - Pronoun inversion
  - Echolalia phase longer
  - Inappropriate use of questions
  - Ego-centric language
  - Later develoment of « I »

Slower and more progressive development
but often few differences at school age.
Alternative pathway.
Verbal language important for compensation of
Difficulties linkes to lack of visual input.

Galiano, Portalier et al (2012); Perez-Pereira & Conti-Ramsden (2002)

Challenges in the perception of sensory information

Peu documentés dans la recherche, observations cliniques. Caffier & Panien (2017).

- Fear of certain noises especially if sudden or out of the child's control
- Taste sensitivities : trouble accepting new textures or tastes..
- Sensory seeking, especially for vestibular or auditory stimuli
- Touch aversion for certain objects or textures (cold, soft, sticky etc.)

Atypical play behaviours

Hatwell (2003) ; Erwin (1993) ; Brambring et Tröster (1992) ; Dale et al (2017)

- Babies « too calm »: less play activity
- Less exploratory behaviours than sigted peers
- Favour solitary, repetitive play or sensory play
- Less symbolic or pretend play
- Less interested in group games

Difficulties with social interactions

Peterson et al (2000); Bigelow (1995, 2003); Dale et al (2013) ; Baron-Cohen (1995) ; Perez-Pereira (2005); Greenaway & Dale, (2017)

- Less frequent joint attention
- Delay in understanding and correctly answering in tests of false belief
- Delay in dveloping theaory of mind
- Rigidity in social interactions
- Emotional withdrawal
- Difficulties integrating with peer groups

DSM-V (2013) /ICD 11 (2022)

ASD

"Dyad" of symptôms

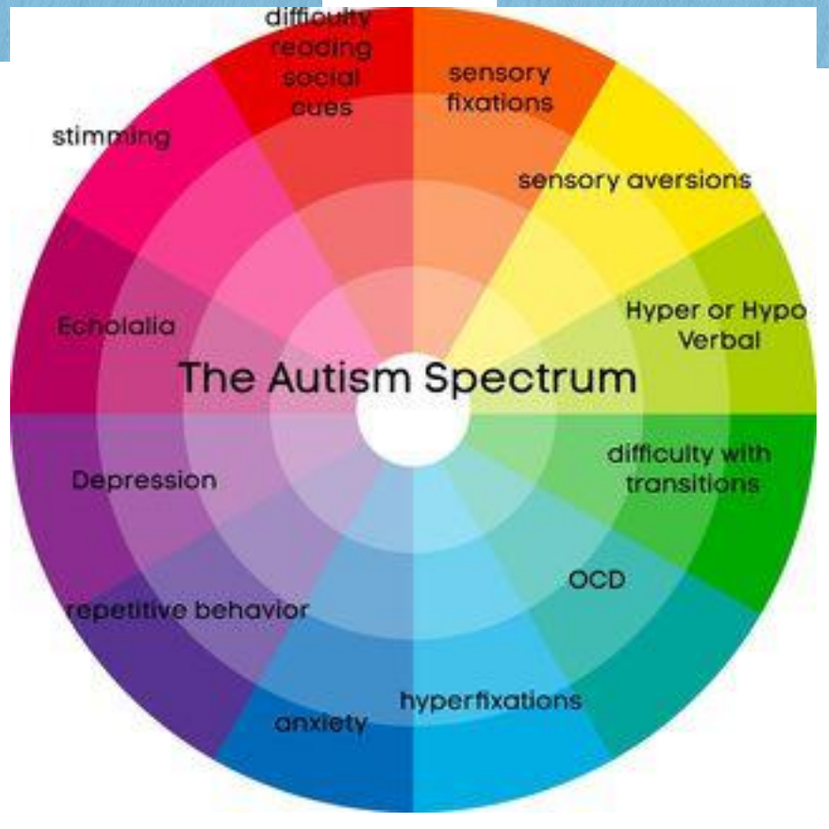
Qualitative alterations of social communication

Repetitive and restricted interests and behaviours

Unusual sensitivity to sensory information

"Spectrum" of difficulties

Variable intensity, comorbid learning disability or not, variation of profiles.



Early detection criteria ASD

M-CHAT-R (Robins, Fein & Barton, 2009) – **target behaviours or skills** in children aged 16-30 months

- ***Visual joint attention**: *The child looks when an adult points to a toy in the room*
- ***Response to name call (expressed differently in VI child)**
- ***Pointing to request something.**
- ***Presence of motor stereotypies**
- **Sharing:** *The child brings something to show to the adult*
- ***Social smiling (tends to occur in reaction to other, non-visual stimuli such as adult closeness or physical play)**
- ***Eye contact**
- ***Interest towards peers**
- **Sensitivity to noise**
- ***Imitation**
- ***Seeking adult's attention (again, shows differently in VI children).**
- **Understanding instructions.**
- ***Symbolic play.**

*** Aspect potentially affected by a visual impairment**

VI-ASD ?

VI+ASD – Not a new topic !

1749 : Diderot – language and cognitive delays in blind people as well as the « absence of emotions »

1914 : Villey – warns parents of blind children against the over-assisting their children, so as to avoid them developping « selfish and ungrateful » attitudes.

1932 : French – observes intense sensory seeking in blind children, obsessions

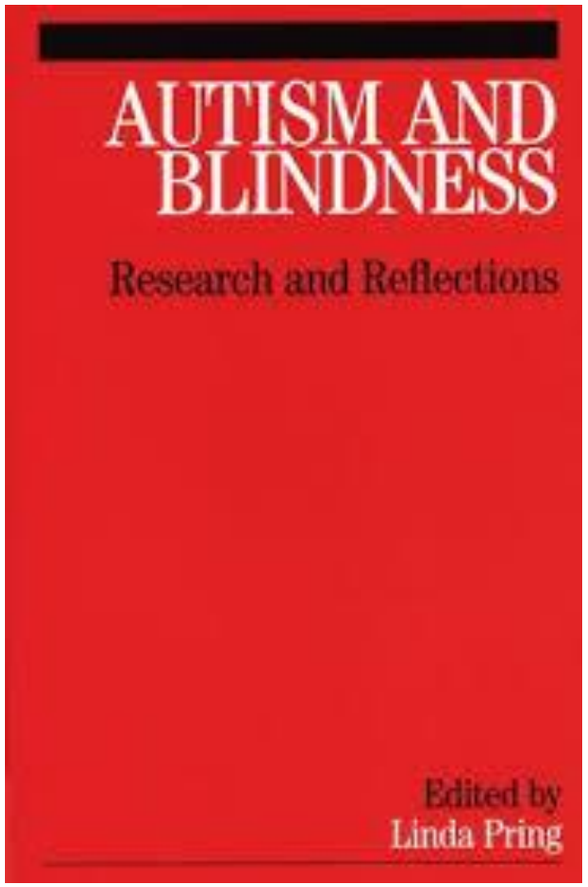
1933 :Cutsforth – emotional instability, daydreaming, sensory seeking.

1942 : Fulcher – lack of appropriate emotional facial expressions, trouble establishing emotional relationships

1958 : Henri – tendancy towards self centredness and emotional withdrawal.

Many authors from the **1950s onwards** mention autism like troubles…ex Keeler

Autism and Blindness…



Autism and Blindness : literature review

Ed. Linda Pring (2005)

Reynolds & Culican (2023) Visual Autism. Children (Basel).

De Vaan et al (2018) Assessing ASD in people with sensory impairments combined with intellectual disabilities. J Dev Phys Disabil

Petrotto el al (2023) Children with ASD and severe VI : Some general principles for intervention according to the clinical psychology of disability. J Public health Res.

Chokron et al (2020) The inter-relationships between CVI, ASD and ID. Neuroscience and Biobehavioural reviews.

Ludwig et al (2021) Considerations for the differential diagnosis of ASD in medically complicated pediatric populations. Clin Neuropsychol.

What the research says…

Autistics TRAITS in blind or visually impaired individuals…

Motor stereotypies and“blindismes”

Fazzi et al (1999)
Smith et al, (1969)
Tröster et al (1991)
McHugh & Lieberman (2003

Verbal communciation charcteristics

Fraiberg (1977)
Fazzi et al (2007)

Atypical repetitive Play

Hatwell (2003)
Erwin (1993)
Brambring et Tröster (1992
Dale et al (2017)

Difficulties in social interactions

Peterson et al (2000)
Bigelow (1995, 2003)
Dale et al (2013)

Full ASD SYNDROME (meets diagnostic criteria (ICD 10/DSM IV or V)

1% *(Pry, 2014)*
3% *(Jan 1977)*
12% *(Mukkades 2007)*
25% *(Tadic 2009)*
30 à 50% (*Parr, 2010; Hobson & Lee, 2010; Absoud, 2011; Jutley-Neilson, 2013; Ek, 1998)*
72% (*Jure , 2016)*

“Autism-like”…but different ou temporary ?

Pry (2014) : Multiple developmental delays in language, motor skills and social skills may mime autism spectrum disorder in VI children

Qualitative differences in ASD symptoms?
Brown et al (1997)

Some symptom diminution or « recovery » in later childhood
Hobson & Lee (2010); Williams (2014) ; Jure (2016)

Some compensation through language development
Perez-Pereira & Conti-Ramsden (1999); Galiano, Portalier et al (2012)

“What we are observing may only be superficial similarities”.
Baron-Cohen (2002)

“A behaviour which appears identitical may have a different function for a blind child and a sighted autistic child”. Perez-Pereira & Conti-Ramsden (2013)

Clinical differences IV – ASD+IV

Despite these similarities: other authors observe qualitative differences *(Salt, 2010)*

Comparison tables (clinical observations – very little research)

Gense & Gense (2005), Brandsborg et al (2012), Pawletko & Rocissano (2000)

LANGUAGE	
ASD+VI	**VI**
Very little verbal language Long period of echolalia Little interest for verbal input Few spoken interactions Repetitive themes	Shorter echolalia phase Takes plesure in verbal interactions Extensive use of language, wide vocabulary

INTERACTIONS SOCIALES	
ASD + VI	**DV**
Little interest in social interactions Little or no sharing behaviours Solitary play Little social engagment	Lots of social curiosity Asks many questions Shares emotions, thoughts and ideas, interesting finds etc.,

Multiple and multifactorial (genetic, environmental), heterogeneous pathogenisis ?

Social communication disorder as a direct consequence of the absence of visual input and the resulting difficulties in establishing relational synchrony

Comorbid neurological lesions, or a genetic syndrome affecting both vision and social communciation.

The combination of total congenital blindness, a neurological susceptiblity and a fragile social-emotional context

ASD = One SYNDROME, many etiologies

Developmental trajectories of visually impaired children

Often, typical development in the first year

Critical period : middle to end of second year : 3 risk groups

Sonksen & Dale (2002); Dale & Salt (2008)

Clear regression In language and social skills “Developmental setback”	Uneven development + Autistic traits	Good communication but rigid with poor flexibility

During childhood and adolescence 2 trajectories observed :

1. **COMPENSATION – Child récuperates social communciation skills at least partially. Autism symptoms decline.**

2. **INSTALLATION – Clinical picture of ASD persists or worsens**

Hobson & Lee (2010); Dale & Salt (2008 ; Jure (2016)…

Problem :

How to detect during early childhood which children are at risk of developping severe social communication disorders ?

= the level of **impact** of such disorders on the **developpement**, future **independance** and **la quality of life of the child** and their family

= Adapting type and i**ntensity** of interventions offered to children and families

Actually 2 problems

1. The difficulty in getting a valid standardised assessment of deviation from the norm in the development of visually impaired children. Few tests available.

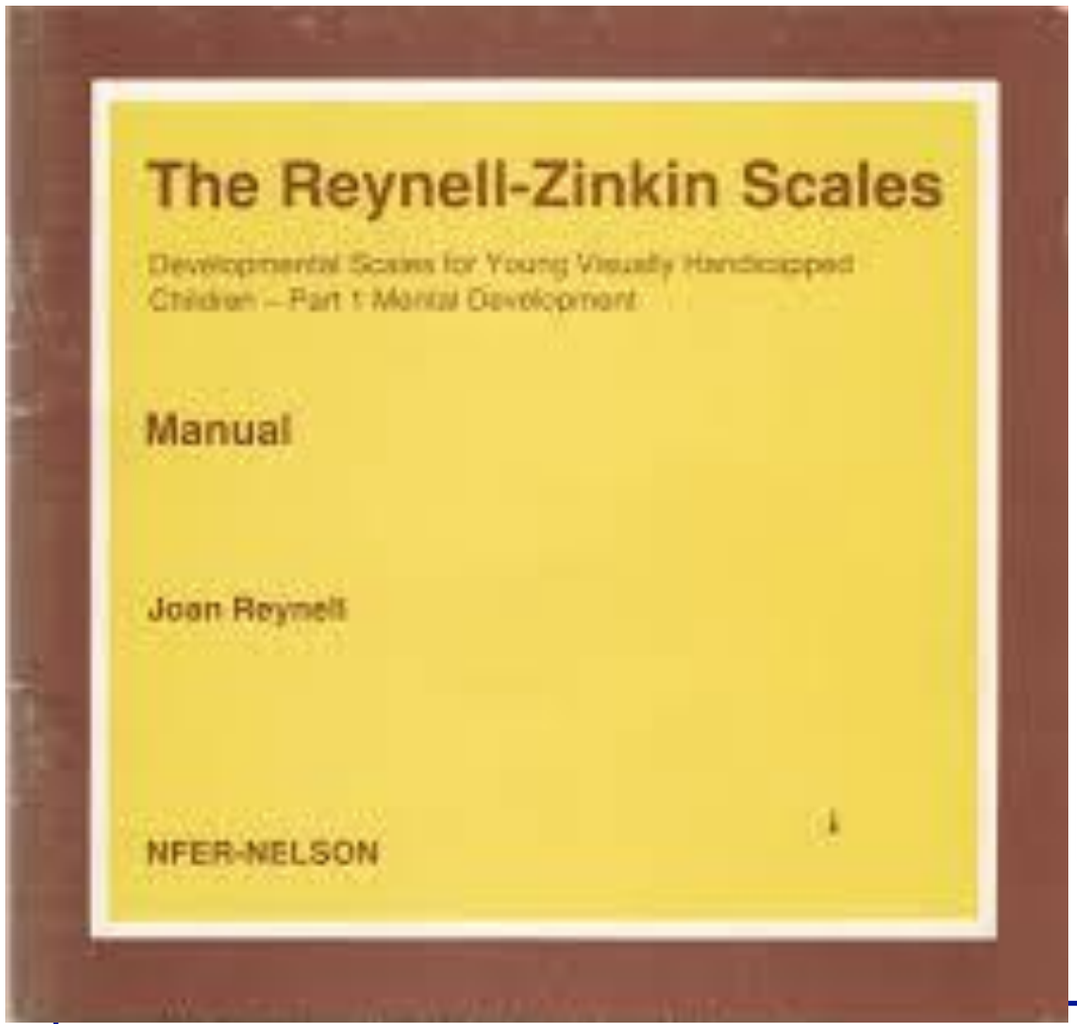
2. The overlap in clinical symptoms which makes diagnosis of ASD difficult in the blind or visually impaired child.

1. 1. The difficulty in getting a valid standardised assessment of deviation from the norm in the development of visually impaired children. Few tests available.

- Lack of specialised assessment tools for VI children.

- Heterogeneity of the VI population (which probably explains the lack of tests !)

The Reynell-Zinkin Developmental Scales for Young Visually Handicapped Children



Developmental assessment- specific to VI children aged 2 to 6 years

Blind / visually impaired (standardised on two sample groups)

6 areas of development (play and exploration, motor, cognitive, language, social…)

Not translated into French
Not standardised with a French cohort group
Edited et standardised in 1979 (Netherlands sample revised in 2000 - Vervloed et al, 2000)

Work in progress…

Project EVAL-DV
Réseau AURA TSA DS

Project GROSJEDI
CNRHR/GAPAS Loos les Lille

2. The overlap in clinical symptoms which makes diagnosis of ASD difficult in the blind or visually impaired child : 2 risks

OVERDIAGNOSIS

Similarities of the clinical signs = ASD is seen in all of the blind or severely VI children due to developmental differences.
“they all have ASD”

DIAGNOSTIC MASKING

Autism spectrum disorders are obscured by the pre-existing VI and are seen as a mere conséquence of the visual impairment, that will pass as the child grows up.
“*its not really, real autism*”

IMPORTANCE OF RELIABLE INFORMATION FOR CLINICAL DECISION MAKING

Pilot study:

Latour, L. (2019). Étude rétrospective des signes précoces des troubles du spectre de l'autisme chez de très jeunes enfants déficients visuels : étude exploratoire. A.N.A.E., 159, 225-234.

RESEARCH QUESTION:

Can we retrospectively identify at a very early age (12 to 48 months) observable signs of autism spectrum disorder in visually impaired children who were later diagnosed with ASD ?

Hypothèses :

1. There will be observable early differences in the levels of social communciation

…less joint attention, less social interest, shared fun, social engagement

2. The signs of autism in early infancy will be specific and clinically different to behaviours linked to adaptations to visual impairment.

…less play, more stereotypies, more sensory sensitivities, atypical language, repetitive movement

Methodologie

Restrospective, comparative study
Home movies

6 infants

Bilateral blindness (WHO level 4/5)

Group ASD VI		Group VI	
3 children, ASD diagnosis		3 children, no developmental problems	
Fille	Leber's Amaurosis	Garçon	Norrie's syndrome
Fille	Genetic syndrome (chromosomic)	Fille	Bilateral Retinoblastoma
Garçon	Optic Nerve Hypoplasia	Garçon	Septo-optic dysplasia

Methodologie

14 films
180 seconds each

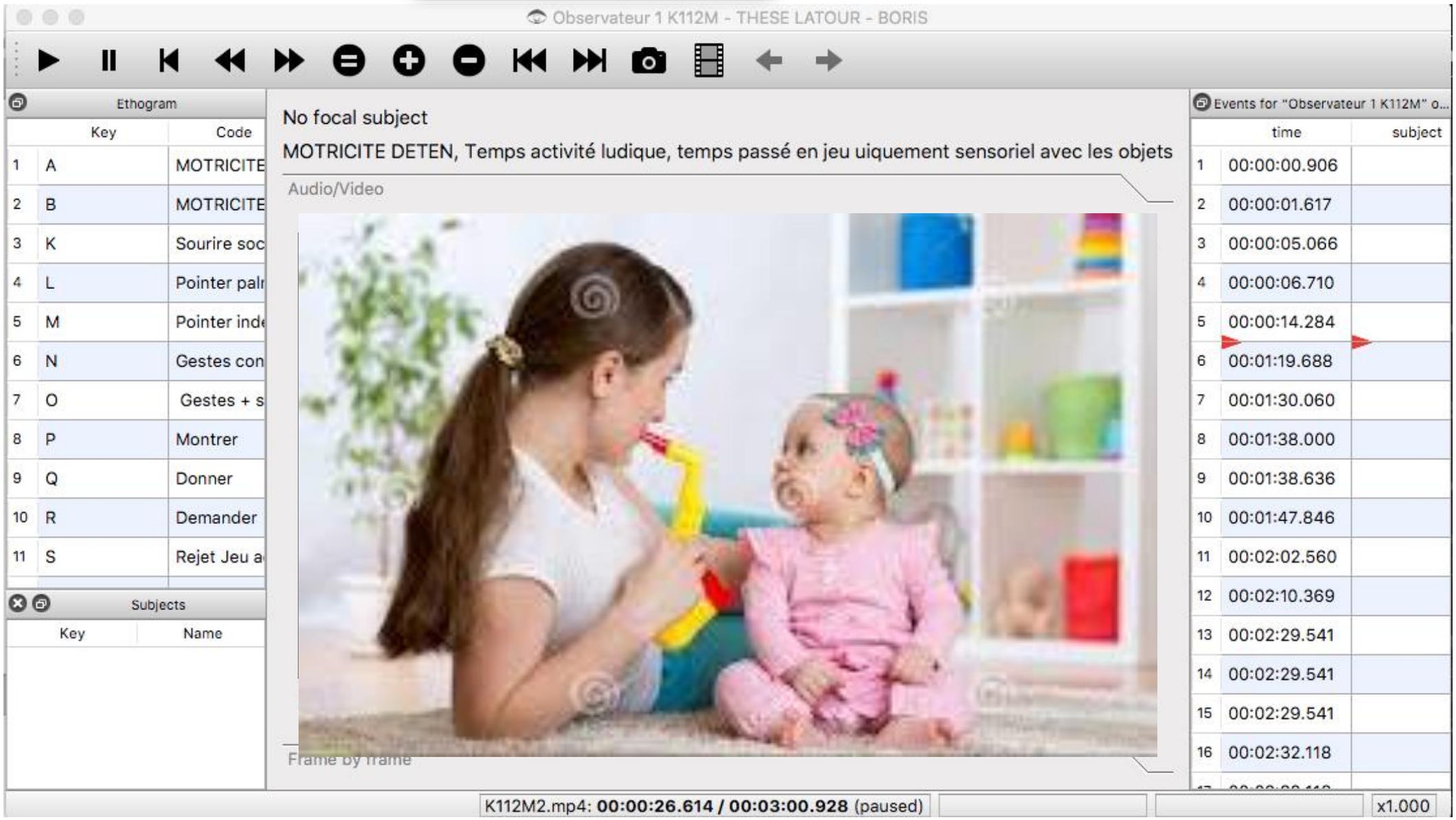
Situations : play and interaction

Film sequences :
12 months - 4 films
24 months - 6 films
3 - 4 yrs - 4 films

Analysed with programme BORIS

CATEGORIES OF BEHAVIOURS OBSERVED
Motor skills
Social Communication and Interactions
Joint attention
Play behaviours
Repetitive Behaviours
Sensory sensitivities
Language and Vocalisations

2 observers



Behaviours coded by 2 observers, the second observer unaware of the child's pathology or group.

Inter-judge fidelity average of 0,96.

Results

Descriptive statistics

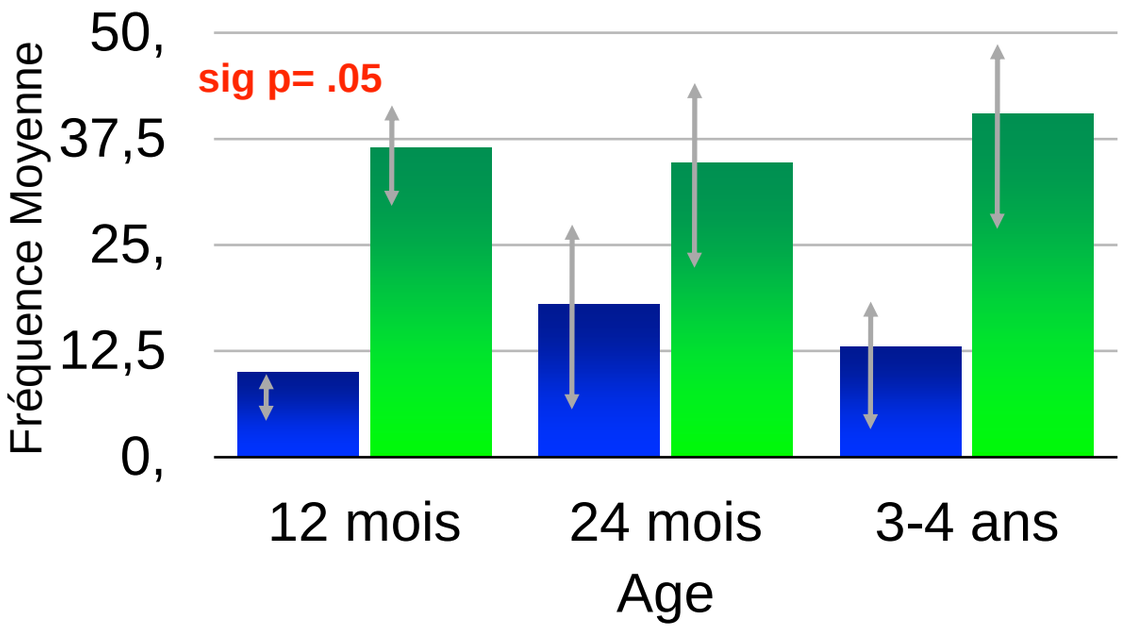
Inferential statistics – transformed data+ test t student

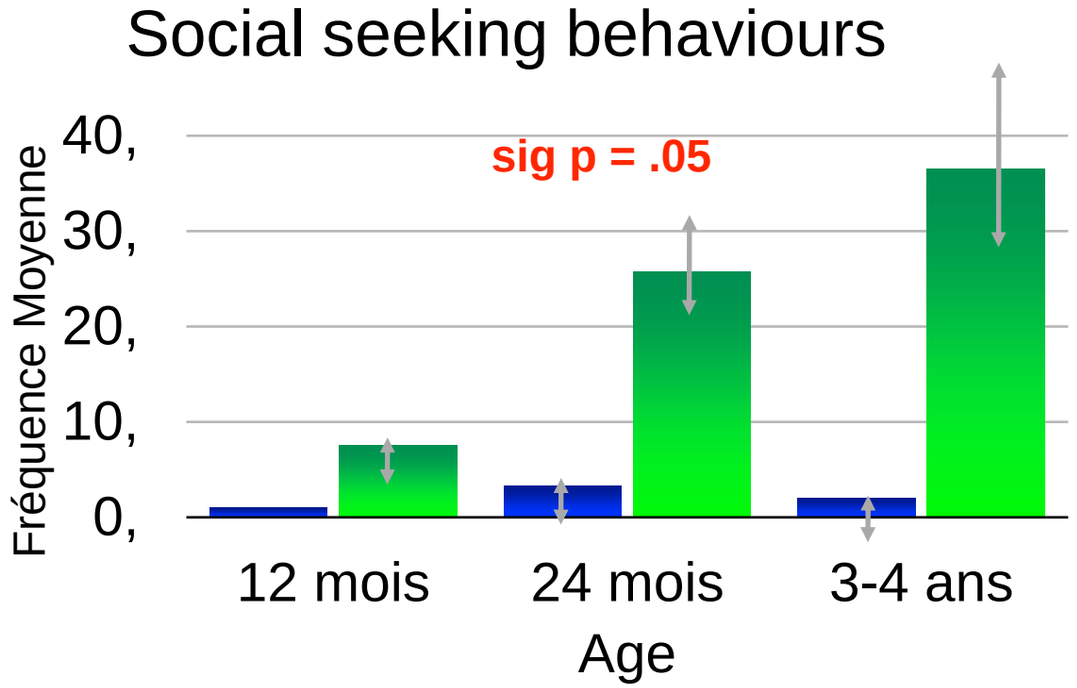
Pilot study, small sample = tendancies
(Poor statistical strength – no generalisation possible)

- **JOINT ATTENTION**
- **SHARED FUN IN SOCIAL INTERACTIONS**

  Some observed differences, but large inter-subject variabilité

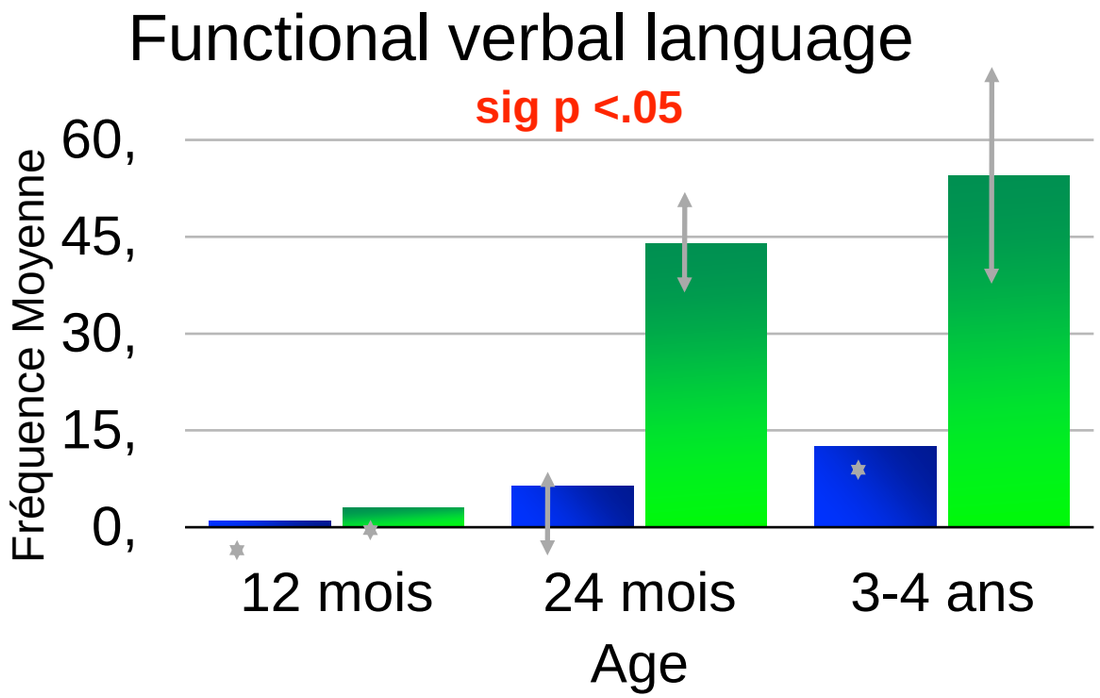
Social communication and Interactions



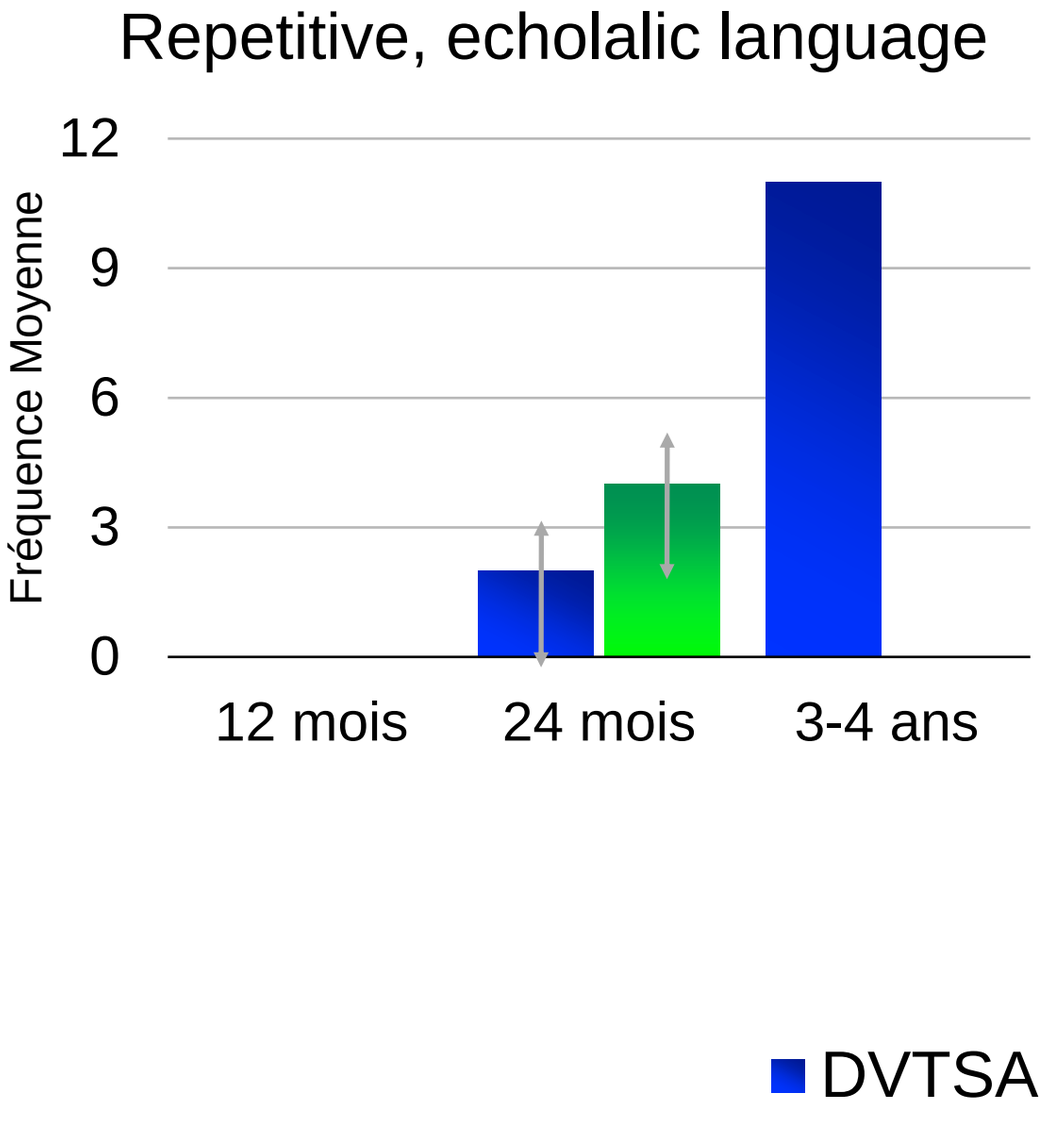


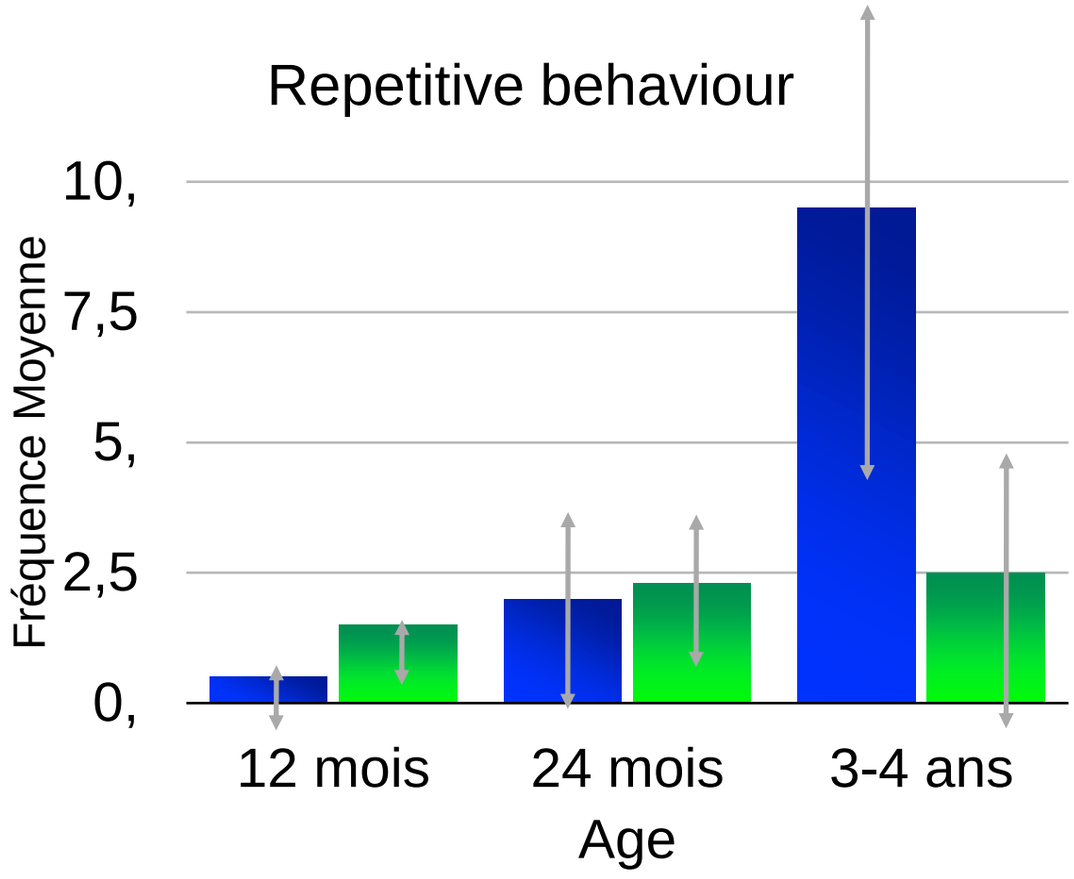
Clear differences observed between the two groups As early as 12 mois /24 mois

Different trajectories Between the two groups



Behaviours not distinct before 3 ans

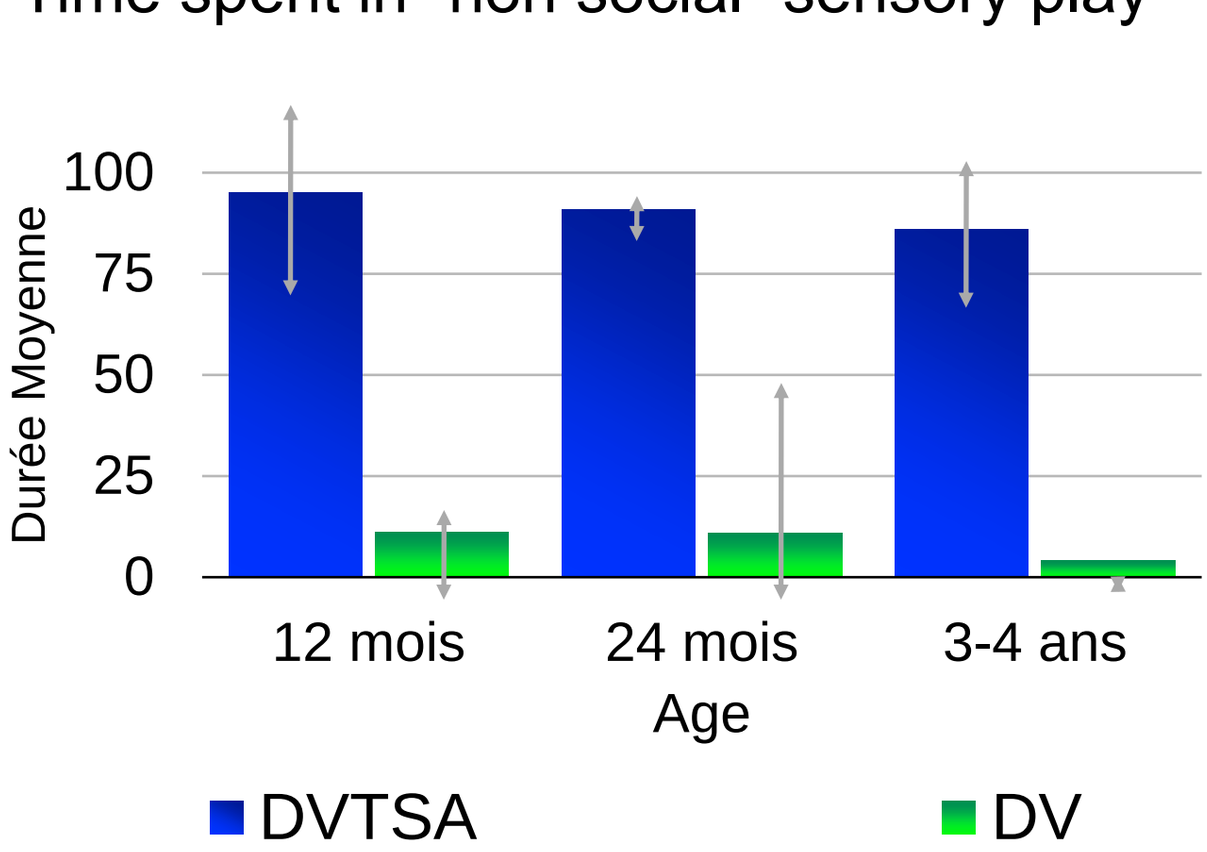




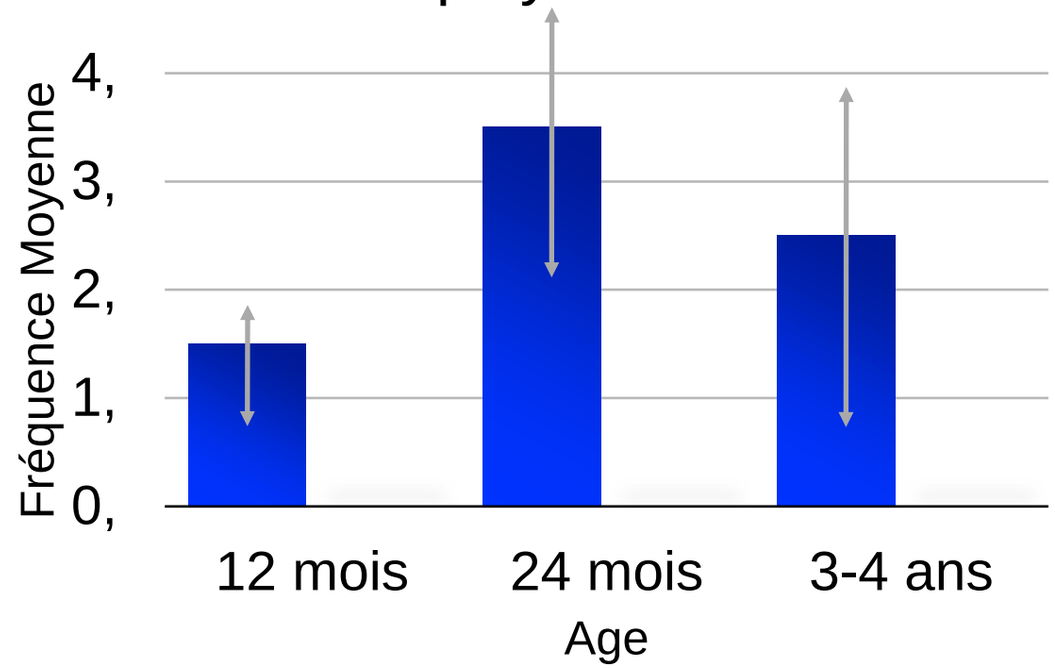
DVTSA DV

Aspects especially (or only) observed in the ASD-VI group, seem interesting for the differential diagnosis

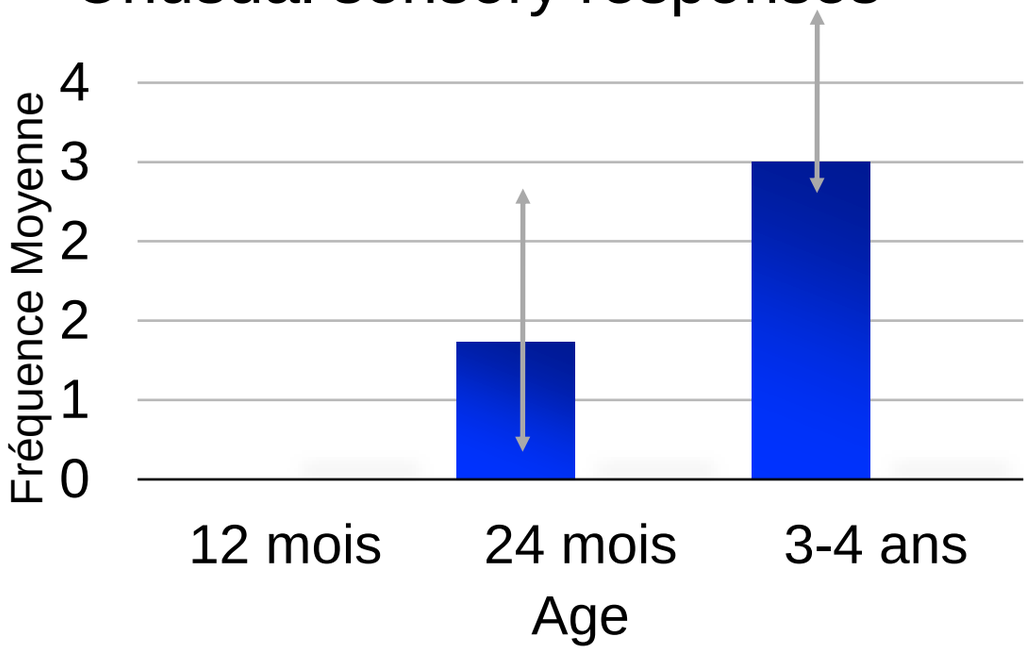
Time spent in “non social” sensory play



Rejecting an adult or peer’s offer of play



Unusual sensory responses



Poor discriminators under the age of 3 :

Symptom overlap ++

- **Stereotypies movements**
- **Absence of response to name**
- **Lack of apparent social interest**
- **Poor imagination**
- **Absence of pointing**
- **Absence of conventional gestes**
- **Repetitive language like echolalia**
- **Absence of symbolic play**

M-CHAT-R	ADOS-2
• Visual joint attention • Response to name • Pointing to ask or show • Motor stereotypies • Sharing • Social smile • Eye contact • Interest for peers Noise sensitivty • Imitation • Seeking adults' attention • Understanding instructions • Symbolic play	• Pointing • Spontaneous directed vocalisation • Gestures for communication • Directed facial expressions • Combining eye contact and requesting • Asking • Joint Attention: initiated by the child or in response to adult • Quantity and quality of social exchanges • Showing or giving an object • Motor stereotypies and repetitive interests

Adaptations needed !!

Seem useful for early detection of ASD in VI children

12 months	24 months	3-4 years
Rejecting an adult's offer of play		
Little manifestations of joint attention		
Few moments of social engagement/ few initiation of interactions		
Long moments of play spent in non social sensory seeking activity		
Not many vocalisations directed to others		
Lack of facial expressions	**Sensory sensitivities (sensory fascination or avoidance)**	
Less shared joy	**Absence of sharing behaviours such as showing**	
	Very little functional language	
	Moments of physical tension	
		Absence of functional play
		Absence de symbolic play
		Increase in atypical language (écholalia, stéréotypies)
		Increase in motor stéréotypies

Conclusions

This study identifies certain behaviours observable at a very young age, which may be useful for detecting ASD in VI children. Need to replicate with a larger cohort or plan a longitudinal study (more control over video content to improve method)

Suggests real possibilties of early detection of ASD despite the symptom overlap

Highllights the need for specific tools, adapted to VI population

<u>TOOLS CURRENTLY IN PREPARATION – DIFFERENTIAL DIAGNOSIS:</u>

- **VISCOS** (Visual Impairment Social Communication Observation Scale). *Sakkalou (2015)*
- **SOCI-VI** (Social Communication Interview for Young Children with Visual Impairment). *Dale, Tadic & Sonksen (2014)*

TOOLS AVAILABLE

- **OASID (Observation of autism in people with sensory and intellectual disabilities)** ***De Vaan et al (2018)***

OASID, an Instrument for Assessing Autism Spectrum Disorders in Individuals with Intellectual Disabilities Combined with Visual Impairments or Deafblindness

de Vaan, G., & Vervloed, M. P. J. (2021). OASID, an Instrument for Assessing Autism Spectrum Disorders in Individuals with Intellectual Disabilities Combined with Visual Impairments or Deafblindness. Journal of Visual Impairment & Blindness, 115(2), 134–142. https://doi.org/10.1177/0145482X211000965

TOOLS AVAILABLE

TEATSA®

TEST POUR ENFANTS AVEUGLES
AVEC TROUBLES DU SPECTRE AUTISTI
OU TROUBLES APPARENTÉS

Functional assessment of blind children (not originally designed as a diagnosis tool)

Cognitive, sensory and communication skills assessed

Child’s competencies and difficulties = project and realistic aims

Non standardised. Validated by a scientific committee of experts.

Adaptation of tools

- Theurel et Gentaz (2014) : Some tools have been specifically developped for the assessment of certain aspects in VI children, but the standardisation and validity of these tests may be criticized.

- Non VI tests : assessment with materials, instructions and cotations not adapted :
  - Vineland (Bechla, 2016) : 29% of the *Socialisation* questions are linked to visual capacities
  - ADI – use discussed by Curran and Fitzpatrick (problems of interpretation)
  - ADOS-2 : Joint attention : « look » + pointing, imitation etc.
    - Several teams have proposed changes to the materials : ex Curran & Fitzpatrick Child Vision Dublin/ synodia Lyon/ Jutley
    - But no consensus as yet
    - Interpretation of results – +ve cotation does not always = asd diag
  - *WISC – verbal subtests, but some questions biased against VI kids : ex : vocabulary : define the word « transparent » !!*

Research questions

QUESTIONS ON THE UNDERLYING ISSUES :

- Is there a special link between Autism and Visual Impairment (especially congenital blindness) ?

  Jure 2019 : Brain changes linked to Superior Colliculus ? But : not 100% ASD

CLINICAL QUESTIONS

- What does Autism Spectrum Disorder “look like” in children and adults with VI ? During the early years ? During the teenage years ?

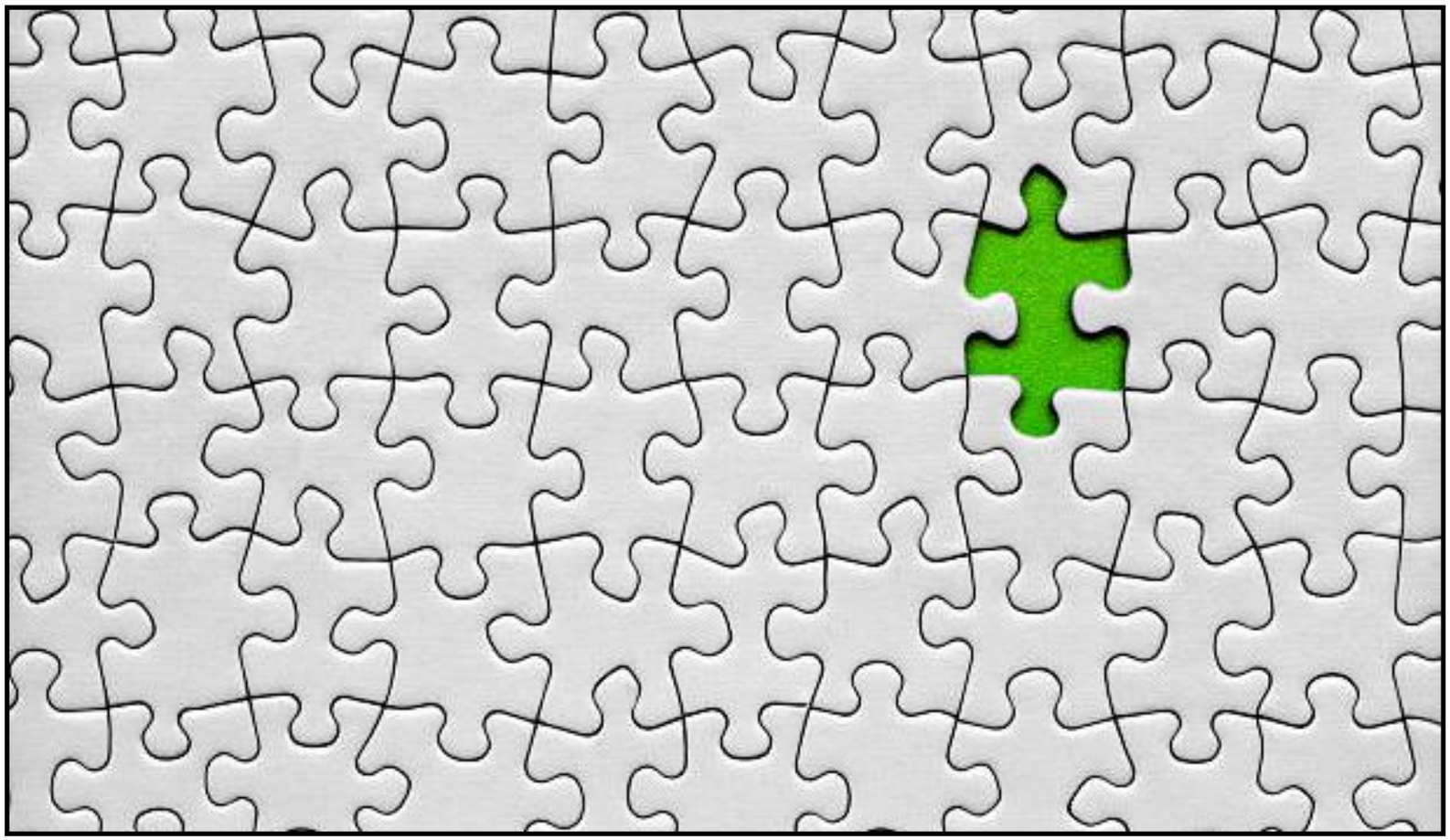
- How can a diagnosis be reliably made ? Which assessments are valid for use with VI children ? Can a formal protocole be agreed on ?
- What adaptations can/should be made to the “gold standard” tests(ADOS/ADI ?)

- What specific tests exist for this group of children or adolescents ?

- Should an ASD diagnosis in a VI child always be considered « temporary »?

- How should care and educational intervention be adapted to children with this complex profile ? Techniques from autism interventions / techniques from visual impairment programmes ?

AUTISM SPECTRUM DISORDERS & VI



Thankyou for your attention