



Children with Angelman syndrome understanding and guiding behaviour

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In close cooperation with the

ENCORE-team

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Angelman Syndrome expertise center



- Optimizing care for children with Angelman syndrome (AS) and their families
- Integration of research, behaviour and learning (neuroscience, neurology, genetics, pediatrics, psychology, psychiatry)
- Understanding mechanisms behind behavior and learning in AS



Behaviour in children with Angelman syndrome

Clinical diagnostic criteria

In 100%

- Developmental delay
- A lot of smiling, happy appearance
- Stereotypies: hand flapping, mouthing
- Hyperactive behaviour
- Language expression < comprehension of language, pictures and gestures

In 20-80%

- Problematic sleeping pattern, feeding problems
- Increased sensitivity for heat
- Strong preference for water, glistening and crackling objects



NB: No known difference in behaviour between genetic subtypes

(Summers et al. 1995, Summers & Feldman 1999, Clarke & Marston 2000, Walz & Benson 2002, Oliver et al. 2002, Chidden et al. 2004, 2006, Barry et al. 2005, Honster & Oliver 2006, Walz 2006, Pelc et al. 2008)

Understanding behaviour in children with AS

- You have several children in-one:
 - Just your child, with a name, parents, a home, aged ...
 - The child with a mental age of ...
 - The child with (features of) Angelman Syndrome
 - The child with features of ASD/ADHD/SMD
 - ...



Guiding behaviour starts with explaining and understanding

- What happened?

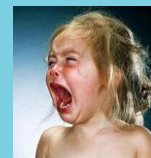
What part of my child plays a role?

- Physical issues?
- History?
- Mental age?
- Real age?
- Communication/language issues?
- ASD or ADHD?
- Sensory issues?
- Stress or fatigue?
- Is it yourself? Parents are human too!



1. What happened?

- Little sister is crying, next to your kid with AS
- Normal child-rearing issues
- Don't let common explanations dominate the scene
 - He is always...
 - Of course she did it...



1. What happened?

- Antecedents:
 - Children ask for clearness and predictability
 - Learn to ignore- pick your battles
- Consequences:
 - Don't let common explanations dominate the scene:
 - He is always...
 - Of course she did it...
 - Consistency in response (don't feel guilty if you are not!)



2. Physical issues

- So 'normal' that you sometimes forget...
- Hunger/low sugar level
- Thirst
- Sleep/fatigue
- Illness (coming or going)
- Pain:
 - Aspecific reactions
 - Location
 - Constipation?



2. Physical issues

- Regularity in:
 - Drinking
 - Feeding
 - Sleep/Rest
- Find a doctor that knows AS!
- Medication (for stool, pain, sleep, epilepsy)



3. History

- Fear, trauma and reliving experiences can be part of life
- Fear can be part of personality (careful?)
- Trauma relives with objects, persons (animals) and situations
- Attack is part of defense



3. History

- Know what happens/ happened
- Explain your child to others
- Anxiety:
 - Don't ignore- keep control and keep contact!
 - Distraction
 - Use language (Were you scared? Was the dog fast? Did it frighten you? That was a big dog!)
 - In phobic or post-traumatic anxiety: use EMDR



4. Mental age

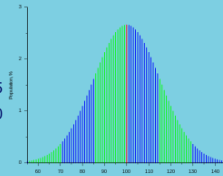
- Is your child a baby, a toddler, a kindergarten-kid?
- Next to that: what is his/her best/worst ability?
- Developmental tasks:
 - Baby : growing trust and attachment (womb space)
 - Toddler : independence with safety (parent space)
 - Kindergarten : independence in exploring limits (kid power)



4. Mental age: Developmental level and IQ

- Intellectual disability, classification DSM IV-TR

- Average : 85-110
- Borderline : 71-84
- Mild : 50-55 tot 70
- Moderate : 35-40 tot 50-55
- Severe : 20-25 tot 35-40
- Profound : IQ<20-25



- Problem in defining ID: Measuring with an unsuitable test
- Calculating IQ : $100 \times \frac{\text{mental age}}{\text{chronological age}}$,
e.g. Child of 10 with dev. level of a 2-year-old
: $100 \times \frac{2}{10} \Rightarrow$ IQ approximately 20

Development in Angelman syndrome



Cognition (learning) is more than intelligence:

- Research from 2004: developmental top 17 months (20 children up to 10 years old)
- Research from 2010: average dev. age 40.5 months (92 children up to 5 years old)
- Strong correlation cognition and adaptive development
- Children with gene-deletion stronger delay in all domains (but language)

Specific profile:

Cognition > language and motor development
 Language comprehension > language expression
 And: relatively strong short term memory
 visual spatial skills ↓↓
 use of objects > imitation

5. Real age



- Physical and mental development can be separated
 - Children compare themselves, but have different rules for different children
 - Skill-development happens unpredictable, keep on looking for just-right challenges!
- Every day get back to your Patience and your Humor!
(Unlearning takes time)
- Respect sexual feelings and acts, but teach where and when



6. Communication and language

- Less communication- more withdrawal or aggression

- Contact: eye, touch- I see/hear you!
- Shared attention
- Pointing
- Choosing objects
- Choosing pictures
- Sentences
- Stories



6. Communication and language

- Look for a Speech-language therapist with experience in intellectual disabilities
- Greenspan; DIR Floortime/ Floorplay : interaction is the key
- PECS: picture exchange communication system <http://www.pecs.com/>
- ABA: applied behavior analysis (e.g. Functional analysis and functional communication training; Radstaake, 2012)
- PRT: Pivotal Response Treatment
- Computer-speech-systems



7 A. Autism spectrum disorders (ASD) in Angelman syndrome

Core symptoms:

limitations in reciprocal social contact, communication and behaviour/play

Considerations:

- Interest in contact and communication
- Is laughing appropriate for the situation?
- Distinguish from low social developmental level
- Overlapping features (language, stereotypes, fascinations)
- AS children do better socially than same-level-non-AS children with ASD
- Variable estimations of percentage of autism



Studies with high incidence:
 63% (10/16; Trillingsgaard & Ostergaard 2004) 42% (8/19; Peters et al. 2004)
 Studies with low incidence:
 <1% (Cohen et al. 2005, Veltman et al. 2005, Smith et al. 1996, Saitoh et al. 1994)

7B. ADHD in Angelman syndrome

- Core symptoms ADHD: **impulsivity, inattention, hyperactivity**
- Impulsivity** frequent (Barry et al. 2005)
- Short **attention** span and distractibility part of normal behaviour in toddlers
- Attention span increases with age (Clayton-Smith 2001)
- Attention can be affected by AED (anti-epileptic drugs)
- Hyperactivity** is part of AS (100%)
- Hyperactivity decreases with age
- Little research in treatment of hyperactivity



Aggression in AS



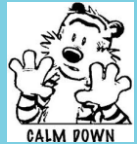
- Is it aggression?
- Or is it: boredom, overstimulation, anxiety, communication??
- Seeking contact, stimulation, rest, comfort, shelter?
 - Give it! On *your* conditions
- Golden rule: Hurt no-one, Destroy nothing-
- Medication
 - Hyperactivity/inattention: stimulant medication (e.g. Methylphenidate)
 - Aggressive behavior/hyper-arousal: neuroleptics (e.g. Risperidone)

7&8. ASD, ADHD and SMD: Behaviour with a function



- (Stereotypical) behaviour can have a reason:
 - Boredom : Self-stimulation to self-mutilation
 - Stress : soothing, calming behaviours
 - What works? What does it bring you?
 - Old behaviour : behaviours that once *had* a reason
 - Social learning : who reacts/reacted?
 - Attracting attention : depends on what happened before

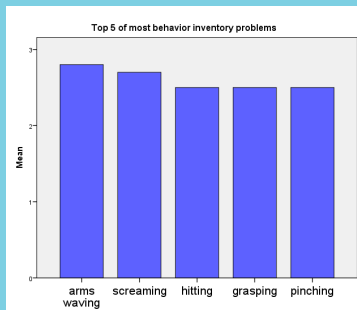
8. Sensory modulation disorders



- Self-regulation in children with low threshold:
 - Calming down
 - Avoiding
 - Using which sense? (e.g. auditory, tactile, mouth,...)
- Self-regulation in children with high threshold:
 - Seeking for stimuli and alertness
 - Seeking for challenges?
 - Using which sense? (e.g. auditory, tactile, movement,...)

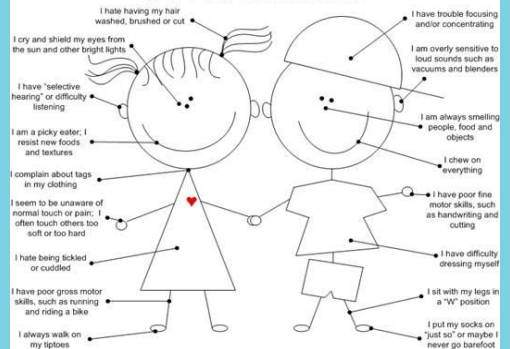


Behavior Problems Inventory



(Rogahn et al. 2001)

DO YOU KNOW ME?



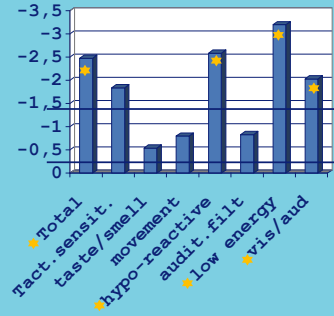
8. Sensory Issues

Problems in sensory processing

- Stimuli from 6 senses:
 - Touch, movement, taste, smell, visual and auditory systems
 - Can be admitted/ conducted (Low threshold-sensitization)
 - Can be inhibited/stopped (High threshold- habituation)
- So stimuli can be experienced as being:
 - Absent, too weak or
 - Very present/ too strong

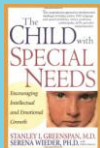
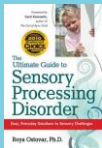


SSP



Sensory processing in AS

- Instruments: Sensory profile, SPM, OTY evaluation
- Walz and Baranek, 2006, N= 340:
 - A lot of sensory processing issues
 - Especially hypo-responsivity (high threshold) for tactile and movement (vestibular) stimuli
 - Independent of sex, epilepsy or genetic subtype
 - Possibly 'sensation seeking behaviour'



9. Stress and fatigue

- Stress and fatigue make *problems* worse and *solutions* harder to find
- Who is tired/stressed out: you or your child?
- Stress in ASS = physical status + sensory stimuli + emotions + demands
- Low stress = health + balance + control + good thinking = A LOT!
- Take care of yourself:
 - Take turns
 - Get rest, relax
 - Do (nice) things together
 - Use your network



9. Sleep problems (in AS)

- Sleep problems are frequent in AS *children*, fatigue in AS *parents*
- Sleep-hygiene:
 - Before sleep:
 - Physical fatigue
 - Light and melatonin (accumulation?)
 - Darkness: screens
 - Starting to sleep:
 - Bedtime (6 o'clock + age/4)
 - Darkness: curtains and bed-lights
 - Rituals
 - Waking up:
 - Predictability
 - Ignoring
 - Safety

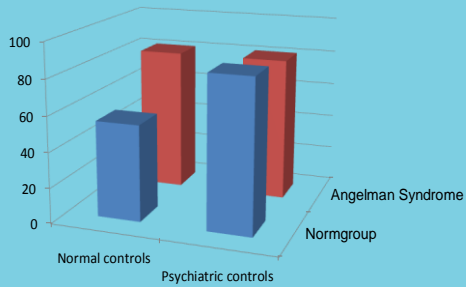


10. Good-enough parenting

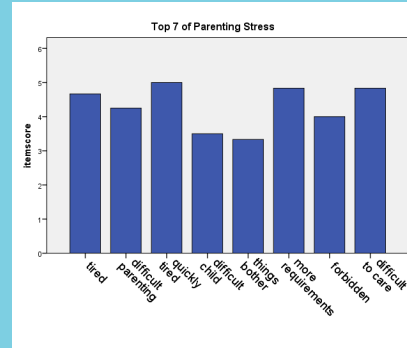
- It is alright to be angry, as long as you don't lose control
- Losing your temper: get out of the situation!
- Be nice to yourself and your colleague/love:
 - quality time is not only *with* your child(ren)
- Angelman parents are SUPER parents!



10. Is it yourself? Parents are human too! (Parental stress index)



PSI: Parental Stress Index



It's the parent's fault

- Parent-blaming
- Good-enough parenting
- Most children know that (grand)parents differ
- Losing** your patience: get out of the situation!
- Be nice to yourself and your colleague/love
- Angelman parents are SUPER parents!



Who helps?



- Pediatrician
- Pediatric neurologist
- Physician for people with intellectual disabilities
- Psychologist
- Psychiatrist
- Social worker
- Speech language therapist
- Physical therapist
- Occupational therapist

WHAT helps?

- Ask yourself why?
- Prevent boredom, overstimulation and other stressors
- React predictable:
 - Ignore** what can be ignored when only attention is the goal
 - Interrupt:** Say, gesture or 'look' NO
 - Prevent** continuation of the behaviour
 - Say** and **show** what you want: (Don't pull- be nice!)
 - Reward** positive behaviour
 - Remove** out of the situation



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Neurofibromatosis 1, Tuberous Sclerosis, Angelman Syndrome