Children with Angelman syndrome
understanding and guiding behaviour

André Rietman, neuropsychologist
In close cooperation with the ENCORE-team
Erasmus Medical Center
Rotterdam
The Netherlands

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

Understanding behaviour in children with AS
- You have several children in one:
  1. Just your child, with a name, parents, a home, aged …
  2. The child with a mental age of …
  3. The child with (features of) Angelman Syndrome
  4. The child with features of ASD/ADHD/SMD
  5. …

Guiding behaviour starts with explaining and understanding
1. What happened?
   What part of my child plays a role?
2. Physical issues?
3. History?
4. Mental age?
5. Real age?
6. Communication/language issues?
7. ASD or ADHD?
8. Sensory issues?
9. Stress or fatigue?
10. 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?

- 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

- 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

- Problem in defining ID: Measuring with an unsuitable test
- Calculating IQ: 100 x mental age/chronological age, e.g. Child of 10 with dev. level of a 2-year-old: 100 x 2/10 => 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
  1. Contact: eye, touch: I see/hear you!
  2. Shared attention
  3. Picturing
  4. Choosing objects
  5. Choosing pictures
  6. Words
  7. Sentences
  8. Stories

- Look for a Speech-language therapist with experience in intellectual disabilities
- Greenspan; DIR Floortime/ Floorplay : interaction is the key
- 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/ploy

Considerations:
- Interest in contact and communication
- Is laughing appropriate for the situation?
- Distinguish from low social developmental level
- Overlapping features (language, stereotypies, 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:
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 behavior 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) behavior can have a reason:
  - Boredom: Self-stimulation to self-mutilation
  - Stress: soothing, calming behaviors
  - What works? What does it bring you?
  - Old behavior: behaviors 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,...)

8. Sensory Issues

Behavior Problems Inventory

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

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)

![Graph showing parental stress index comparison between Normgroup, Angelman Syndrome, and Psychiatric controls.]

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

ENCORE: Expertise center NeuroCognitive Developmental disorders Erasmus Rotterdam

Neuroscience
Prof. Dr. Ype Elgersma

General Paediatrics
Prof. Dr. Henriette Moll
Karen de Haas, MD

Clinical Genetics
Dr. Alice Brooks, MD

Pediatric Neurology
Dr. Marie Claire de Wit, MD
Andre Rietman, MS

Child and adolescent psychiatry/psychology
Leontine ten Hoopen, MD
Gwen Deleman, MD

Speech language therapy
Cindy Naves, MA
Physical therapy

Erasmus MC – Sophia Children’s Hospital, Rotterdam
Neurofibromatosis 1, Tuberous Sclerosis, Angelman Syndrome