

Down Syndrome & Fragile-X Syndrome



solution-s

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Terry Broda

RN[EC], BScN, NP-PHC, CDDN

What is DS?

- A common genetic variation (3 genetic ways)
- So certain genes on chromosome 21 are “overexpressed” & this usually causes health problems & intellectual & developmental disabilities (I/DD)
- Exact causes currently unknown
- Most common cause of I/DD
- Not related to race, nationality, religion or socio-economic status.

What is it?

- Incidence: about one in 700-900 live births
- Likelihood of giving birth to a child w/ DS increases with maternal age

BUT:

- 80% of bbs w/ DS are born to women <35yrs (because women <35yrs give birth to more babies overall!).
- Wide variation in I/DD, behavior & physical development. Each has his/her own unique personality, capabilities & talents!

How?

3 genetic ways:

- 95% have trisomy 21 (an extra chromosome 21 in all their cells),
- 3-4% have a translocation form of the extra chromosome (where the extra chromosome 21 is attached to one of a different chromosome pair)
- about 1-2% are mosaic (only some cells are trisomic, the rest are normal)

Physical features

- Upward slanting eyes w/ epicanthal folds
- Brushfield spots (eyes)
- Flat nasal bridge
- Simean crease (hands)
- Clinodactyly (hands)
- Short stature
- Small ears & mouth
- Protruding tongue w/ high arched palate

Physical Health

- Hypothyroidism (by age 50: 40%)
- Epilepsy (6-13%, but 50% have adult onset by 50yrs, & if DS + dementia, it can be as high as 80%!)
- Ortho:
 - Atlanto-axial instability
 - Degenerative disc disease of C-spine
 - Hip disease (28%)
- Gastrointestinal issues:
 - GI tract abnormalities at birth (8-12%) (duodenal stenosis or atresia, imperforate anus, Hirschsprung disease)
 - Celiac disease
 - Constipation, GERD, H. Pylori

Physical Health

- Sensory Issues:
 - Hearing deficits (66-89%)
 - Higher risk AOM/OE
 - Visual problems (60%)
(strabismus, keratoconus, cataracts)
- Skin conditions (50%): eczema, dry skin
- Early onset menopause (44.6yrs)
- Cancers:
 - Leukemias (10-30X more common in childhood)
 - Testicular (25% have undescended testes)

Physical Health

- Obesity (50-60%)
- Dental concerns (60-100%):
 - gingivitis, bruxism, malocclusion
- Respiratory Issues:
 - Obstructive sleep apnea (OSA)
 - Higher risk of pneumonia & URTI
- Cardiac issues:

(17% in previously undiagnosed adults, of which 25% needed semi-urgent care; Vis&al)

 - Congenital heart defects (30-50%)
 - Mitral valve prolapse
- Life expectancy: 45-55 yrs old but they can even live into their 90s now!

Mental Health

- Depression (6-11%, & higher levels if they have dementia, too)
- OCD :obsessional slowness & “the groove”
- GAD (anxiety)
- ASD, ADHD
- Self-talk (81%!): typical or a sign of mental health issues: psychosis, depression or anxiety?
- Early-onset Alzheimer’s dementia (>40yrs: 15-45%)

Depression?

Changes in:

- Behavior (irritability, listless, paranoia, decrease in skills (ADLs), more self-talk)
- Appetite
- Sleep patterns
- Activity level
- Interactions: passivity, withdrawal & mutism
- Changes in memory?

DM-ID, (2007), p.30-32.
NDSC website:
www.ndsccenter.org/?page_id=778

Differentials?

- Sleep apnea
- Hypothyroidism
- Vitamin B12 deficiency
- Depression
- Cerumen impaction
- Hearing loss
- Dementia: early screening recommended
- Pain!

DS – strategies vs. Challenging behaviours

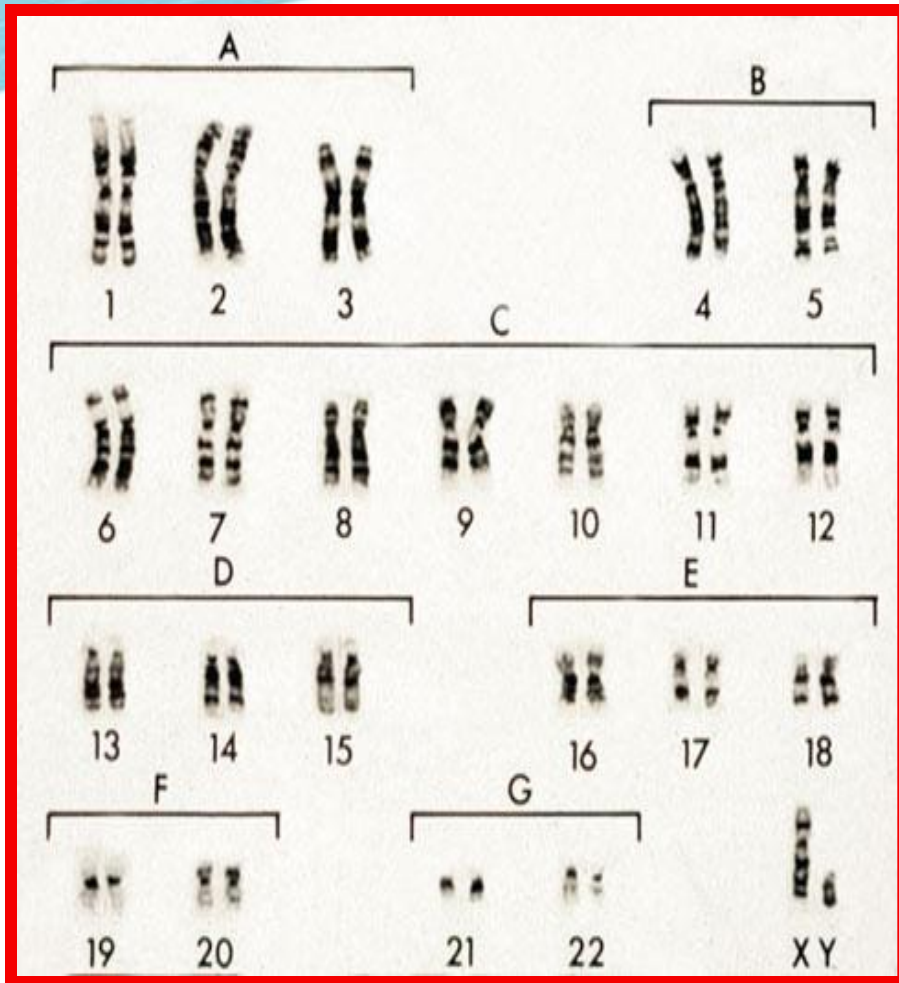
- **Functional analysis: A-B-C data collection**
- “Stubbornness” or “running away” may be due to fear or avoidance: need to identify the underlying purpose of the behaviour!
- May also be d/t underlying medical problem: always rule out the physical!
- Scrapbook & adaptations to environment may be helpful to the person with dementia

Interventions

- Early intervention is best: OT, Speech referrals
- Work together with the families
- Have families also work with the school
- Respite & community supports!
- See handouts on websites
- Also DSE website for educational software:
<http://www.down-syndrome.org/practice/350/>

Fragile X Syndrome

Fragile –X Syndrome



- Section **q27.3** of the **X** chromosome
- Prevalence:
 - 1 / 1500-4000 males
 - 1 / 2500-8000 females
 - 1 / 256 females are carriers of the premutation
(Rousseau et al., 1995)
 - 1/800 males are carriers
- Present in all ethnic groups

Fragile –X Syndrome

- Most common inherited cause of DD
- Females usually have milder symptoms (compensation by other X chromosome)
- Often initial dx of autism or PDD-NOS
 - *39% of males with fragile χ had dx of autism or PDD in childhood*
 - *16-17% of adults with fragile χ meet DSM criteria for autism*
 - *0-16% males with dx of autism test + for fragile χ*

Fragile X Syndrome

- Fragile X syndrome is associated with an expanded repetition of the trinucleotide CGG which, in « normal » persons, is repeated between 6 and 50 times.
 - 1) normal = 6 - 50 CGG repeats
 - 2) premutation = 50-200 CGG repeats (*FXTAS)
 - 3) full mutation = 200 + CGG repeats (Fragile X)

*increased impact over generations

*FXTAS: Fragile X-associated Tremor/Ataxia Syndrome

- Progressive neurological disorder:
 - tremor & ataxia (& eventually memory problems, moodiness & irritability)
- Onset 50-60yrs (granddads of Fragile X kids)
- ONLY 20-30% of male carriers >50 affected
- Often misdx'ed as atypical Parkinson's, multiple system atrophy, etc
- May provide insight into FMR1 gene deactivation

Characteristic features:

- Long face
- Prominent chin
- Prominent ears
- Larger head circumference
- Joint hypermobility/hyperextension
- Macro-orchidism

Characteristic features:

- Associated medical concerns:
 - Strabismus
 - Flat feet
 - Mitral valve prolapse (MVP)
 - Obs Sleep apnea (OSA)
 - Autistic features (poor eye contact, hand-flapping, hand-biting)
 - Serous otitis
 - Dislocated hips
 - GERD
 - ADHD
 - Seizures
 - Mental retardation

Shyness, social anxiety & hypersensitivity

- Shy, timid personality
- Difficulties w/ peer interactions compared to interactions w/ adults.
- Excessive anxiety in new situations/environments.
- Hypersensitivity: Tendancy to 'overreact' to 'minor' frustrations .

Sensory Issues:

- **hyper arousal** (sound of fluorescent lights, sight of too many decorations on wall)
- **hypo arousal** (sound of the teacher's voice, rather than the sound of the humming computer, fluorescent lights, and aquarium bubbler)
- **sensory motor integration problems** (including motor planning issues and fine motor weaknesses)
- **tactile defensiveness** (hypersensitivity to touch)
- **difficulty in many new, confusing, or loud situations** (because of a combination of sensory integration problems, anxiety, and attention deficit disorders)

Tactile Defensiveness

- Affects 60-90% FXS boys & some FXS girls
- Overreaction to touch & may avoid it
- Increased or decreased reactions to textures:
 - Clothing & tags
 - Need soft fabrics, no elastic cuffs or hems
 - May prefer deep pressure of heavy clothing for increased feedback
- May have difficulty identifying objects or feeling & receiving info by touch

Tactile Defensiveness

- Firm, sure touches (handshakes & bearhugs) may be tolerated better than light touch (tickling, soft touch of face)
- May prefer to be at end of the line, separate from crowd
- Infants may/may not be comforted by cuddling
- May not enjoy finger painting or other tactile art activities

Tactile Defensiveness

- Difficulties with hygiene:
 - Bathing, face & hair washing, shaving, nail cutting
- Dental visits may be difficult & anxiety provoking
- Difficulties with eating:
 - Difficulty nursing from breast or bottle
 - Strong food preferences related to textures of food
 - Mouth stuffing of mouth, due to high “cathedral” palate, before realizing they may gag

Remember:

- A specific problem in the environment that can be modified will often effect a much larger improvement in behavior than medication!
- Maximize environment FIRST to get a reasonable baseline!

Tactile Defensiveness: strategies for intervention

- Sensory diet: individualized by an OT
- Uses neurodevelopmental therapy working with muscle tone & sensory integration therapy (SI), involving all senses plus proprioception (body position in space) & vestibular (sense of gravity & motion) input
- To find best combination & timing of various sensory inputs & decreases sensory overload

Tactile Defensiveness: strategies for intervention

- Calming activities:
 - Rocking, swinging child
 - Applying deep pressure
 - Brushing child's skin with therapeutic brush
 - Break time: quieter area, playing computer game or listening to music or a story on headphones

Tactile Defensiveness: strategies for intervention

- Environmental changes:
 - Increase natural light
 - Limit/avoid exposure to loud situations
 - Gradual desensitization to be able to tolerate more noise
 - Adapted seating to help maintain upright posture with enough feedback:
 - Donut-shaped cushions, foam wedges

FXS – ADHD

- ↓ distractions: study cubicles, desk at front of classroom or in calm area (facing a wall), periods of quiet time, decreased flow of traffic in room, adequate natural lighting & heat, small group instruction, reduced noise
- Seating near an exit, & allow exit PRN
- ↑ use of visual cues (photos) 4 transitions
- Provide non-verbal cues & feedback
- Simple phrases & concrete communication
- Structure/routine/predictability

FXS- general approach

- Do not force eye contact! (gaze aversion)
- Be careful invading personal space & touching the person! (tactile sensitivity)
- Consistency important! (staffing, schedules, environment)
- *Provide a book to carry with them containing info that may be difficult to remember

FXS – ADL stuff

- Sleep:
 - PJs & bedding
 - Dark room/shades
 - Soothing sounds, music
 - Bedtime routine
- Eating:
 - Try various nipples/positions
 - OT interventions for improved oral motor functioning

FXS – ADL stuff

- Dressing:
 - Remove tags, soft fabrics
 - Buttons, snaps easier or T-shirts
 - Shoes w/ velcro, curly laces
- Hygiene:
 - Desensitization to water on skin, calming strategies
 - Pictures of sequence of activities
 - Firm pressure with facecloth vs light strokes
- Dental
 - Egg timer
 - Desensitization: books, visits w/ mom, sibling

*Fragile X Syndrome - issues around sexuality

- Social Sexual skills
- Sex Ed. throughout & beyond puberty
- Sexual abuse prevention information
- Psychotherapy & counselling (self-esteem, depression, anxiety/frustration) (especially helpful for transition from parents' home to independent living)

FXS – strategies vs. aggression

- **Functional analysis: A-B-C data collection**
- Aggression may be preceded by giggling, non-compliance or avoidance
- Most common cause: anxiety!
- May be d/t sensory processing problems or hypersensitivity: sensory stimulation 'adds up' during the day & sensory activities may be more challenging later in the day (↑ demands are more difficult)
- *higher incidence in adolescents: hormones!



FXS - strategies vs. aggression

- Indications of anxiety:
 - Worry/anxiety over changes in routine or upcoming stressful events (fire drills, assemblies): 'hyper vigilance'
 - Will stiffen up when angry or upset, becoming rigid & tense
 - May simply tighten up hands
 - Crying, whining tantrums may all be d/t overwhelming settings!

FXS – strategies vs. aggression

- “Catch them being good!” with reinforcement of + behavior
- Specific interventional approaches: ABA, Lovaas, token economy, ‘time-outs’ (removal of attention)
- Psychotherapy & individual counselling (self-esteem, depression, anxiety, coping skills, frustration, anger management, social skills)
- Family Therapy

FXS- strategies vs. aggression

- Relaxation training, sensory stimulation/sensory integration (OT), music
- Deep pressure massage
- Use of imagery
- Group Therapy & Social Skills training (role playing, especially with behavioral consequences)

FXS – strategies vs. aggression

Consider differentials:

1. **Panic episodes: 'fight or flight':** flushing, turning pale, rapid breathing, sudden sweating
2. **Mood disorders: Depression or Bipolar disorder** (disturbed or absent sleep, excess or loss of appetite, changes in activity level, mood changes, increased irritability)
3. **Seizure disorder** (aggression appears aimless, or unassociated with any ongoing event, occurring with unusual movements, brief loss of consciousness, confusion or need to sleep afterwards)

Websites

Health Watch tables for several genetic syndromes & other tools from the Canadian Consensus Guidelines developed at Surrey Place in Ontario, Canada:

www.surreyplace.on.ca/Clinical-Programs/Medical-Services/Pages/PrimaryCare.aspx

FXS Websites

- The Fragile X Research Foundation of Canada
Website: <http://www.fragile-x.ca>
- FRAXA Research Foundation (USA)
Website: www.fraxa.org
- The National Fragile X Foundation (USA)
Website: <http://www.nfxf.org/html/what.htm> or
<http://www.fragilex.org/html/home.shtml>

DS References

- Prasher,V.P., Asim, F., Holder, R (2004). *The Adaptive Behaviour Dementia Questionnaire (ABDQ) : Screening Questionnaire for Dementia in Alzheimer's disease in Adults with Down syndrome: Research in Developmental Disabilities*, 25(4):385-97.
- DMR dementia screening tool, see attached.

DS Websites (Cdn)

- Canadian Down Syndrome Society Website: www.cdss.ca/
- Down Syndrome Research Foundation (Canada) website: www.dsrf.org
- Down Syndrome Association of Ontario website: www.dsao.ca/
- Down Syndrome Association - National Capital Region website: www.dsancr.com/

DS Websites: (USA & UK)

- National Down Syndrome Congress Website:
www.ndsccenter.org
- National Down Syndrome Society
Website: www.ndss.org
- National Association for Down Syndrome Website:
www.nads.org
- Down Syndrome Association-UK
Website: www.dsa-uk.com
- Down Syndrome Education
Website: www.down-syndrome.org
- International Mosaic Down Syndrome Association
Website: www.imdsa.org

Genetics Websites

- Geneclinics : <http://geneclinics.org/> (see: Gene Reviews)
- Your Genes, Your Health: <http://www.ygyh.org/>
- Online Mendelian Inheritance in Man: <http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM>

Genetics Websites

- Genetics Education Center, University of Kansas Medical Center:
<http://www.kumc.edu/gec/support/>
-
- The Family Village:
<http://www.familyvillage.wisc.edu>
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Websites

Genetics Website

(In English, French, Spanish, German, Italian & Portuguese!):

- Orphanet: <http://www.orpha.net/consor/cgi-bin/index.php>



solution-s

29-2450 Lancaster

Ottawa, Ontario K1B 5N3

T 613 249-8593

F 613 249-0198

info@solution-s.ca

www.solution-s.ca