Down Syndrome & Dual Diagnosis

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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)

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%)

Differentials?

- Sleep apnea
- Hypothyroidism
- Vitamin B12 deficiency
- Depression
- Cerumen impaction
- Hearing loss
- Dementia
- Pain

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

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?

References

- www.ds-health.com
DS: monitoring

- [http://www.denison.edu/collaborations/dsq/recordsheet.html](http://www.denison.edu/collaborations/dsq/recordsheet.html)

References

- DMR dementia screening tool, see attached.

Websites (Cdn)

- Canadian Down Syndrome Society Website: [www.cdss.ca/](http://www.cdss.ca/)
- Down Syndrome Research Foundation (Canada) website: [www.dsrfg.org](http://www.dsrfg.org)
- Down Syndrome Association of Ontario website: [www.dsao.ca/](http://www.dsao.ca/)

Websites (USA & UK)

- National Down Syndrome Congress Website: [www.ndsccenter.org](http://www.ndsccenter.org)
- National Down Syndrome Society Website: [www.ndss.org](http://www.ndss.org)
- National Association for Down Syndrome Website: [www.nads.org](http://www.nads.org)
- Down Syndrome Association-UK Website: [www.dsa-uk.com](http://www.dsa-uk.com)

Additional Website

- International Mosaic Down Syndrome Association website: [www.imdsa.org](http://www.imdsa.org)