

SOLUTION S

## Certification Preparation: Seizures & Syndromes

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## Other syndromes & conditions

- Autism
- Spina bifida
- Cerebral Palsy
- Fetal alcohol syndrome
- Congenital rubella
- Tourette syndrome

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- **Keys to success**
- **Overview of Syndromes:**
  - Down
  - Fragile X
  - Williams
  - Prader-Willi
  - Angelman
  - Smith-Magenis
  - Cornelia de Lange
  - Rett
  - Tuberous Sclerosis
  - Velocardiofacial (22q-)

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## Down Syndrome

- 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.

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## \*\*Keys to success

- Etiology
- Clinical characteristics/symptoms:
  - Physical features
  - Medical co-morbidities
  - Cognitive & behavioral aspects
- Natural history
- Management:
  - Treatment interventions
  - Side effects of treatments
  - Intervention strategies

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## 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!

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## 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)

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## 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)

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## 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

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## 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!

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## 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

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## 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%)

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## 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.ndscenter.org?page\\_id=778](http://www.ndscenter.org?page_id=778)

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## 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 X had dx of autism or PDD in childhood
  - 16-17% of adults with fragile X meet DSM criteria for autism
  - 0-16% males with dx of autism test + for fragile X

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## References

- [www.ds-health.com](http://www.ds-health.com)
- Health watch tables, Canadian Consensus Guidelines, Ontario:  
[www.surreyplace.on.ca/Clinical-Programs/Medical-Services/Pages/PrimaryCare.aspx](http://www.surreyplace.on.ca/Clinical-Programs/Medical-Services/Pages/PrimaryCare.aspx)

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## 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)
- An FMR1 gene with a full mutation becomes inactive (methylated) & cannot produce the FMR1 protein (which plays a key role in brain development!).

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## DS: monitoring

- <http://www.denison.edu/collaborations/dsq/recordsheet.html>
- Recordsheet Birth to Age 12:  
<http://www.ds-health.com/recordsheet1.htm>
- Recordsheet Age 13 to Adult:  
<http://www.ds-health.com/recordsheet2.htm>

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## FMR Premutation

**In males & females:**

- FXTAS: Late-onset, progressive cerebellar ataxia & intention tremor (40% risk in premutation males > 50yrs)
- Minor features may include parkinsonism, short term memory loss, executive function deficits.
- Increased risk of ASD & ADD

**In 20% of premutation females:**

- Premature ovarian failure (POF) with cessation of menses < 40 yrs.

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## Fragile X Checklist

Mental retardation  
Hyperactivity  
Short attention span  
\*Tactilely defensive  
Hand-flapping  
Hand-biting  
Poor eye contact  
\*Perseverative speech  
\*Hyperextensible MP joints  
Large or prominent ears  
Large testicles  
\*Simian crease or Sydney line  
Family history of mental retardation

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## FXS: monitoring

- GERD
- Connective tissue: pes planus, hyperflexibility
- Echocardiogram
- Vision & hearing assessments (ophthalmology & ENT)  
Speech & language, OT, PT

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## Characteristic features:

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

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## Behavioural symptoms

- Excessive anxiety in new situations or environments.
- Hypersensitivity: Tendency to 'overreact' to 'minor' frustrations
- Sensory issues: tactile sensitivity

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## Characteristics:

**Associated problems:**

- Strabismus (30-56%)
- Recurrent serous otitis in childhood
- Dislocated hips, Scoliosis, hernias
- ADHD
- Mental retardation
- Autistic features (poor eye contact, hand-flapping, hand-biting)
- Tactile sensitivity
- Hyperextensible joints, flat feet
- Epilepsy (13% -50%)
- Mitral valve prolapse (55%), cardiac murmurs, hypertension
- Hypotonia, poor muscle tone in childhood

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## Shyness, social anxiety & hypersensitivity

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

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## Tactile Defensiveness

- Affects 60-90% of 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
- Have difficulty identifying objects or feeling & receiving info by touch

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## 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

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## 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

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## Tactile Defensiveness: strategies for intervention

- Calming activities:
  - Rocking, swinging child
  - Applying deep pressure
  - Breaktime: quieter area, playing computer game or listening to music or a story on headphones

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## \*\*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!

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## 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

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## Fine & Gross Motor skills

- Movement therapy to improve balance, muscle tone & proprioception:
  - dance, martial arts, sports, physical play
- Practice to improve use of
  - pens/pencils for writing & drawing
  - utensils, scissors & tools
  - Keyboard (computer use)

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## 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

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## Oral-motor activities

- Activities to
  - increase tolerance to touch around face & mouth
  - improve chewing, swallowing & speaking
- Use of foods & toys:
  - Blow toys, whistles, straws
  - Crunchy or chewy food: fruit snacks, celery, bagels, gum
  - \*may decrease chewing on clothing, straps or skin!

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## FXS - ADHD

- ↓ distractions: study cubicles, desk at front of classroom or in calmer area facing a wall, periods of quiet time, decreased flow of traffic in room, adequate lighting & heat
- ↑ use of visual cues (photos, etc)
- simple phrases & concrete communication
- structure/routine/predictability

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## 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

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## Fragile X Syndrome - more teaching approaches.....

- Using pictograms, photos, objects of special interest or hands-on approach
- Using clocks, license plates & cooking to help with number concepts
- Indirect explanation: teach task to neighbour
- Apply person's strengths: long-term memory, imitation skills, sense of humor
- Teach complete tasks: present whole process (not step-by-step) & use cover up method to follow sequence (Ø lose his place)

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### Fragile X Syndrome - general approaches

- 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

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### Strategies vs. aggression

- "Catch them being good!" w/ reinforcement of + behavior
- Specific interventional approaches: ABA, Lovaas, token economy, 'time-outs'
- Psychotherapy & counselling (self-esteem, depression, anxiety\*/frustration, anger management, coping skills, social skills)
- Family Therapy

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### Strategies vs. aggression

- **Functional analysis:**
  - A-B-C data collection
- Aggression may be preceded by giggling, non-compliance or avoidance
- May be d/t sensory processing problems: 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!

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### Strategies vs. aggression

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

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### Strategies vs. aggression

**Consider differentials:**

1. Panic episodes: 'fight or flight'
2. Mood disorders: Depression or Bipolar disorder
3. Seizure disorder
4. Pain due to underlying medical problem

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### FXS- 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)

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## Williams syndrome SOLUTIONS

- Strabismus
- Hyperacusis
- Cardiac malformations: supravalvular aortic (75%) or pulmonic stenosis, HTN, renal artery stenosis
- Renal anomalies w/ frequent UTI
- GI: colic, reflux, constipation, ulcers, diverticuli
- Hernias (umbilical & inguinal)
- Hyperlaxity in youth, contractures w/ age (risk of scoliosis, kyphosis, lordosis)
- Anxiety

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## WS - Intervention Strategies... SOLUTIONS

- Limit distractions: study cubicles, desk at front of classroom or in calmer area facing a wall, periods of quiet time, decreased flow of traffic in room
- Encourage 'talking-through-it' during problem-solving & use of concrete objects/situations (real \$ & real-life situations)
- Encourage use of computers, calculators

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## Williams: monitoring SOLUTIONS

At time of Dx:

- PE & Neuro exam
- Growth chart monitoring
- Consult cardio (echo & Doppler)
- Consult Urology (U/S bladder & kidneys)
- BUN, creatinine & U/A
- Serum calcium & Ca/creat in urine
- TFTs
- Ophthalmology consult
- Developmental exam: speech, cognition

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## WS - Intervention Strategies... SOLUTIONS

- Tape recorded homework instead of written assignments
- Schedule with photos for daily activities
- Digital watches for telling time
- Music therapy/lessons
- Work: people-oriented jobs vs. assembly-line type of work

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## WS: monitoring SOLUTIONS

Lifelong:

- Cardiac: BP X 2 arms annually (HTN risk) monitor for: mitral valve prolapse, aortic insufficiency, arterial stenosis
- Ca+ in serum & urine q. 2 yrs
- \*NO PEDIATRIC VITAMINS for WS kids (vit D)
- Monitor & Tx constipation aggressively!
- Screen adults for sensorineural hearing loss, diabetes & hypothyroidism

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## Intervention Strategies SOLUTIONS

Hyperacusis:

- ear plugs
- quieter environments, limited distractions
- provide warning prior to loud noises (alarms, sharpener, bells)
- comfort the person during their distress
- if room is too loud, leave the area
- \*hyperacusis vs. paranoia!

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## Intervention Strategies...

Anxiety/Fear:

- provide reassurance (but with set limits to ↓ attention-seeking behavior)
- Use distraction by changing topics after initial reassurances
- CBT-cognitive behavioral therapy
- Pharmacotherapy (\*with caution!): start with low doses since WS persons are Rx-sensitive (Ex. Ritalin, tricyclics & ♥)

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## Bristol stool form

- From "Understanding your Bowels" by Dr. Heaton
- Website: [www.familydoctor.co.uk](http://www.familydoctor.co.uk)

\*\*Check out the preview of the book & you'll find the form there

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## WS - Intervention Strategies

Social skills:

- Use 'buddy system' to practice social skills, role-plays, & social stories
- Topics to cover: how to make & keep friends, approaching others, taking turns, conversational skills, dealing with romantic issues & sexuality, how & why to be wary of strangers
- Group therapy can increase self esteem (but may not be appropriate for some d/t anxiety or ADHD)

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## Prader-Willi syndrome

- In infancy
  - Hypotonia ('floppy baby')
  - Feeding difficulties w/ FTT
- Then btwn 1- 6yrs old:
  - Rapid wt gain w/ central obesity
  - Hyperphagia w/ food-seeking, hoarding & foraging behaviors

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## Chronic Constipation

Monitoring:

- Bristol stool form
- Positioning : legs bent at the knee, with thighs elevated to ↑ pressure on the abdomen (crouching) (rocking on the seat)

Interventions:

- Stool softeners to maintain regularity & prevent rectal prolapse

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## PWS-Common characteristics

- Obesity
- Short stature w/ decreased levels of growth hormone
- Small hands & feet
- Almond-shaped eyes
- Hypogonadism

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## PWS

- Obesity-related illness:
  - Diabetes
  - HTN
  - Hyperlipidemia
  - Sleep apnea (ST memory impairment)
  - Pica
  - Cellulitis (skin-picking)

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## PWS - Intervention Strategies...

Food issues:

- \*\*DO NOT USE FOOD AS A REWARD OR PUNISHMENT
- Caloric intake: 800-1200cal/day
- Calcium & vitamin supplements
- Limit access (locks on cupboards, fridge)
- Environmental changes (PWS residences)

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## PWS

- Elevated tolerance to pain (may have injury or # but may not c/o pain)
- Decreased or absent vomit/gag reflex
- Thick viscous saliva: increased caries
- Thermoregulatory instability (masked fever)
- Osteoporosis( increased risk of # ), scoliosis, kyphosis

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## PWS - Intervention Strategies...

Tantrums: (\*verify if due to food issues!!)

- DO NOT ARGUE
- BE CONSISTENT (provide structure & routine)
- set firm limits
- written rules/contracts (with them)
- allow time to settle
- change the environment (quiet area)
- diversion, humour (but not sarcasm)

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## PWS: monitoring

- Developmental & educational assessment along with speech therapy assessment
- Assess males for cryptorchidism
- Strabismus in infants & children
- X-rays to r/o scoliosis
- Bone density to r/o osteoporosis, most likely will need calcium supplementation
- Annual BMI
- HgbA1C for those with significant obesity
- Sleep study to r/o sleep apnea
- Psych assessment to r/o OCD & psychosis

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## PWS - Intervention Strategies...

Skin-picking:

- Reinforce + behavior
- cut nails short
- encourage alternative activities/distractions: collages, scrapbooks, Play-Doh, playing cards, hand cream, puzzles (\*scheduling)
- bandaids may be helpful as an initial layer to be removed before skin-picking can occur (liquid bandaid, long term drsg-duoderm/allevyn)
- HANDWASHING!!!!

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## Intervention Strategies

Educational strategies:

- a.m. classes when person is more alert
- consistency with breaks, lunchtime
- demo's, pictures, models to teach
- specific choices (ex. red or blue crayon not which one)
- encourage helping others
- using puzzles (jigsaw & word search)
- extra physical activities

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## Angelman syndrome

- Epilepsy (86-96%)
- Ataxia (100%)
- Scoliosis (more common w/ advancing age)
- Strabismus (42-75%)
- Sleep problems common
- Severe language impairment  
(expressive impairment > receptive impairment)
- Individuals w/ UPD have less severe ataxia, seizures & language impairments

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## Intervention Strategies

Psychiatric considerations:

- OCD (food rituals - but may also make poor food choices, hoarding, ordering, repeating)
- depression
- Psychosis (more common in UPD)
- ASD (more common in UPD)
- anxiety

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## AS: monitoring

- Baseline MRI & EEG to assess for epilepsy  
(\***AVOID** AEDs that increase brain GABA levels - vigabatrin & tigabine)
- MSK exam to assess ataxia & scoliosis
- Ophthalmology consult to r/o strabismus & assess visual acuity
- Assess for GERD & constipation
- Consults for communication & educational strategies, PT & OT

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## PWS- Strategies

- Growth hormone replacement therapy
- Replacement of sex hormones
- **No medications are known to aid in controlling hyperphagia**

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## AS-Intervention Strategies...

Communication strategies:

- Offer choices (2) & provide item ASAP
- Yes/No cards
- Sign language
- PECS (communication book)

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## AS-Intervention Strategies...

Increase physical activities:

- Hippotherapy
- Hydrotherapy/swimming (\*SUPERVISION)
- ROM exercises
- Therapy ball, floor exercises

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- Vision problems: myopia, strabismus (30-100%)
- Infections: onychotillomania (30%)
- SIB: Polyembolokoilamania (25%)
- ENT problems: otitis & hearing loss, anomalies of the palate (63-81%)
- CV abnormalities: ASD, VSD, valvular stenosis (27-29%)
- Scoliosis (42-65%)
- Renal abnormalities (28-35%)
- Pain/heat tolerance: Peripheral neuropathy (75%)
- "Self-hug" & "lick & flip"

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## Intervention Strategies...

For sleep problems:

- Active day with exercise!
- Establish a routine HS & pm (naps)
- Snack before bed
- Very dark bedroom, dark blinds/curtains
- Weighted quilt/blanket
- Waterbed
- Split-style door
- White noise machine/ fan/ soft music

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## SMS: monitoring

At Dx:

- ROS: PE & Neuro exams
- Renal U/S, Echocardiogram, Spinal x-rays
- Ophthalmology consult, ENT consult, PT, OT
- Speech evaluation, audiology
- Bldwk: immunoglobulins, lipids, TFTs
- Sleep diary & sleep apnea studies PRN

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## Intervention Strategies...

For sleep problems:

- Egg crate mattress, soft blankets
- Warm bath before bed
- Warm milk hs
- Warm H2O bottle on abdomen
- Warm socks!
- Melatonin

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## SMS: monitoring

Annually:

- PT, OT, Speech evaluation
- TFTs
- Fasting lipid profile
- U/A
- Monitor for scoliosis
- Ophthalmology
- ENT: otitis & sinus problems
- Audiology: sensorineural hearing loss

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## SMS: monitoring

PRN assessments:

- EEG (CT, MRI may also be needed)
- Urology W/U if frequent UTIs
- If microdeletion extends into 17p12 then adrenal function must be assessed (SMS = 17p11.2)

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## Intervention Strategies

For aggression & SIB:

- **\*\*CAREFUL W/ PHYSICAL INTERVENTION**
- try to redirect to another activity
- try to determine triggering factors & eliminate them
- applying nail polish may decrease onychotillomania
- Rx: oral contraceptives, SSRI's, mood-stabilizers (AED's)
- \*There has been a relative "calming" noted in adulthood

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## SMS- Intervention Strategies...

Educational strategies:

- sign language, speech therapy
- **structured activities, routines**
- small classes, individual attention (designate them as the teacher's helper)
- computers
- well-matched with teacher (Ø power struggles)
- use creativity, humor

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## SMS – other concerns...

- Tactile defensiveness – may present as stripping of clothes!
- Hugging –can be aggressive: rib-crushing
- Polyembolokoilamania (orifice-stuffing) can include vagina/rectum (vs ? abuse)
- Cavities: large pulp chambers & low levels of enamel
- Peripheral neuropathy (numbness/tingling in fingers & toes) (SIB:hand-biting?)

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## Intervention Strategies...

For sleep problems:

- (\*see AS slides for other interventions)
- Very dark bedroom, dark blinds/curtains
- Weighted quilt/blanket (massaging hands & feet may also help)
- split-style door, enclosed bed (Vail bed)
- SAD lights (pineal gland) (inverted circadian rhythm)
- Melatonin

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## Cornelia de Lange syndrome

- Eye problems: myopia/ptosis/nystagmus (57%), conjunctivitis (67%)
- GI problems (75%); GERD (30%)
- Ear problems (73%); hearing loss (60%)
- Peripheral neuropathy
- Seizures (23%)
- Hernias (hiatal, diaphragmatic, inguinal)
- Ortho: upper limb abN, hip abN (5-10%)

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## CdLS: monitoring



- GERD evaluation, monitor for FTT
- ENT, audiology & ophthalmology
- X-ray upper extremities: radioulnar synostosis
- Monitor for cardiac & renal abnormalities:  
Echocardiogram, renal U/S, vesicoureterogram (VSUG), urologic exam (r/o cryptorchidism & hypospadias)
- Neurology evaluation & EEG
- CBC PRN (bruising, bleeding, anemia suspected)

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## GERD



### Interventions (without an Rx):

- Fill in food diary
- Avoid alcohol, chocolate, coffee, spicy foods, fatty foods, tomatoes, onions, citrus & other acidic fruits
- Smaller meals, more frequently
- Avoid eating 2 hours before going to sleep
- Decrease weight & stop/cut down on smoking
- Raise HOB 6"-8" to sleep (2-3 pillows)
- Observe for vomiting with blood, coffee grains\*

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## Intervention Strategies



### Structure & routine:

- Schedule (with photos or words)

### Communication strategies:

- Pointing
- Yes/No cards
- Sign language
- PECS
- Speech therapy

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## Intervention Strategies



### Tantrums or sleep problems: may be d/t

- GERD
- pain/neuropathy (otitis, conjunctivitis, bunions)
- cold feet (anti-slip socks!)
- Temperature intolerance & altered pain sensation reported by some parents

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## CdLS – Intervention Strategies



### Teach relaxation:

- Music
- Hammock
- Special doll/toy

### Teach ways to decrease agitation:

- Squeezeball
- Deep breathing exercises
- Time out chair/area that can be requested

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## Rett syndrome



- Skin breakdown: hand-mouthing, immobility
- Seizures
- Hyperventilation/breath-holding spells
- Scoliosis (Tx: braces, Sx)
- Constipation
- Poor circulation: cold hands & feet

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## Rett syndrome: monitoring

- Monitor & Tx constipation
- Monitor & Tx GERD
- Consult OT (sensory integration & proprioception, music therapy)
- Consult PT (ROM, hydroTx, orthotics)
- Consult speech therapist for assistive & augmentative communication (AAC) & vs dysphagia & chewing problems
- EEG for sz investigation

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## RS-Intervention Strategies

- Vs skin breakdown d/t hand-mouthing:
  - Hand-holding, distraction/stimulation (favorite toy, keys), massage, gloves/socks
  - Arm/hand/elbow splints PRN (LEAST restrictive!)
- Vs bruxism:
  - jaw massage, orthodontics, teething object
- INDIVIDUALIZED APPROACH!

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## Rett syndrome: Rx

- Vs feeding & growth retardation:
  - caloric supplements
  - Gastrostomy button placement
- Vs seizures: AEDs (but + sensitive, avoid polypharmacy)

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## RS-Intervention Strategies

- Vs hyperventilation, air swallowing, GERD & constipation:
  - No Tx for breath-holding & air swallowing
  - Positioning after meals vs GERD
  - Avoid trigger foods for GERD
  - Monitor stools (frequency & consistency)

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## Intervention Strategies...

### Vs Drooling:

- Flavoured extract applied to oral mucosa & under tongue
- 1% Atropine ophthalmic gtts: 1 gtt orally qd
- Robinol (glycopyrolate) orally, scopolamine patch
- Botox injections orally q.3 months
- bandana, bib with design (for salivation)
- oral stimulation: using a straw, bagels, peanut butter, gum\*
- Surgery: removal of salivary glands, redirection of salivary ducts

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## RS-Intervention Strategies

- Vs sleep problems:
  - Melatonin
- Vs screaming spells:
  - ID causes!
  - Assess for pain (PMS, GERD, etc)
  - Gradual transitions, with parent
  - Music
  - Massage
  - Warm baths

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## RS: Communication

- “speak” to you “through the eyes”
- Eye-pointing behavior
- Comprehension > expression!
- Some have autistic features
- Allow time to express wants
- Parents are excellent resources!
- Immobilization of hands may improve focus on conversation
- Speech Tx: AAC

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## TSC

- Developmental disability
- Autism
- PDD
- OCD
- Anxiety
- Mood disorders
- Chronic sleep disorders

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## Tuberous Sclerosis Complex

- Tubers, subungual fibromas (PAIN!)
- Epilepsy (90%)
- Rhabdomyomata, angiomas (47-67%)
- Renal deterioration & failure (45-81%)
- Ocular issues: hamartoma, astrocytoma (up to 75%)
- Hamartoma that may lead to liver failure (up to 75%)

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## TSC: monitoring

- Abdominal (renal) U/S q.1-3 yrs if abN, CT or MRI
- BUN, creatinine q. 6 months
- Cranial CT/MRI for kids & ados
- EEG for sz F/U
- Chest CT PRN
- ECG initially & echocardiogram PRN
- Ophthalmology consult
- Perform initial exam with Woods lamp to assess dermatological findings

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## TSC

Teeth, skin & nail findings:

- Dental pits (48-100%)
- Gingival fibromas (33%)
- Facial angiofibromas/forehead plaques (77-86%)
- Hypomelanotic macules (“ash-leaf spots”) (3+) (95%)
- Shagreen patches (48-54%)
- Confetti spots
- Ungual fibromas (nails) (11-23% ado; 88% adults)

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## TSC: Rx

- Vs seizures:
  - AEDs
  - Surgery to remove tubers
  - VNS?
- Vs sleep problems: melatonin
- Vs facial angiofibromas: laser Tx

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## VCFS/22q- syndrome

- ENT: Cleft palate, otitis, deafness
- Cardiovascular abnormalities (85%): R aortic arch (52%), Tetralogy of Fallot (21%), ASD, VSD (62%)
- Visual problems: cataracts, tortuosity of retinal vessels (30%)
- Immune deficiency (77%) (\*AVOID live vaccines) (r/t thymus)
- Hyperthyroid
- Hypoparathyroidism w/ hypocalcemia
- Scoliosis, arthritis
- Renal abnormalities: (absent/dysplastic/multicystic kidneys, hypospadias, reflux, obstructive uropathy)
- Mental health issues: anxiety disorders, bipolar disorder, schizophrenia & depression

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## Triggers

- Fatigue
- Exercise
- Hypoglycemia
- Stress (emotional)
- Infection
- Alcohol
- Hyperventilation
- Hormonal changes (catamenial epilepsy)
- Being startled
- Flashing lights

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## Intervention Strategies

- speech therapy (ARTICULATION!)
- Pharyngeal flap Sx possibly
- provide structure
- use + reinforcement
- encourage self-talk/thinking aloud to decrease impulsivity
- computers: to help with reading & spelling, also with math & abstract problem-solving

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## Classification

**PARTIAL SEIZURES:**

- Focal Seizures : the abnormal electrical activity begins in a specific area of the brain ('focus')
- \*By propagation to other portions of the brain, can secondarily generalize

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## EPILEPSY

Seizures – why?

- Genetic – chromosome 22 (Qc), various others including 2,3,6,8,10,16,19,20 & X
- Lesion – birth injury, infection, CVA, tumor
- Post traumatic - head injury (usually 6 months - 2 years after)
- Idiopathic
- Biochemical imbalance (ETOH/Rx OD, E- imbalances)

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## PARTIAL SEIZURES

**SIMPLE PARTIAL:** (NO LOC)

- -can be an 'aura'
- -can be localized in one area of the body (arm, leg, face)
- -can spread further
- -can be motor (movement), sensory (tingling or pain) or visual/auditory/olfactory, & can include sudden sweating, flushing/paleness
- -lasts a few seconds to minutes

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## PARTIAL SEIZURES SOLUTIONS

**COMPLEX PARTIAL:** (WITH LOC)

- usually starts with a blank stare, followed by chewing & some type of random activity
- person may seem dazed & may mumble, or may respond verbally but not appropriately, may repeat the same phrase over & over
- actions can be clumsy & not directed, or can be inappropriate actions ('automatisms': picking at clothing, shuffling papers, or trying to get undressed)

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## Classification SOLUTIONS

**GENERALIZED SEIZURES:**

- BOTH hemispheres of the brain are involved simultaneously, from the start of the seizure
- Consciousness IS impaired

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## PARTIAL SEIZURES SOLUTIONS

**COMPLEX PARTIAL...**

- may wander or run, may appear afraid, may become agitated & struggle or hit if restrained
- usually same pattern occurs for each seizure
- usually preceded by an aura
- lasts 2-5 minutes, but may remain confused & disoriented for ½ hr. or longer

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## GENERALIZED SEIZURES SOLUTIONS

**Atonic** ('drop attacks')

- -loss of muscle tone, person collapses like a rag doll
- -can last from 10 seconds to one minute
- -recovers quickly, person is up and about afterwards

**Myoclonic**

- -sudden, brief muscle jerks of a body part or whole body

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## PARTIAL SEIZURES SOLUTIONS

**COMPLEX PARTIAL...**

- person does not remember what happened during the seizure
- atypical examples:
  - 'GELASTIC' - laughing
  - 'QUIRITARIAN' - crying
  - 'CURSIVE' - running

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## GENERALIZED SEIZURES SOLUTIONS

**Absence**

- -complete loss of awareness, may stare off into space, doesn't answer, & usually doesn't fall down
- -may be accompanied by rapid blinking or some chewing movements
- -unaware of surroundings during the seizure
- -no warning (no aura), short duration (lasts 5 - 30 seconds) & quick recovery (will continue previous activity)

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# GENERALIZED SEIZURES

**Tonic-clonic**

- loss of consciousness, may cry out, falls down, body has jerking movements, lots of saliva, possibly incontinent, may bite tongue
- confusion, fatigue & aggression are possible post-ictally
- can last 2 - 5 minutes

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| Treatment               | Age                | Indication  | Efficacy                                    | Side Effects                                      |
|-------------------------|--------------------|---|---|---|
| <b>AEDs</b>             | Children<br>Adults | Specific AEDs for specific seizure types            | 64% sz freedom (1)                          | Vary by AED, typically CNS- and endocrine-related |
| <b>Ketogenic Diet</b>   | Primarily children | All seizure types                                   | 54% pts >50% sz reduction at 3 months (2)   | Lipid disorders, ketoacidosis                     |
| <b>Epilepsy Surgery</b> | Children<br>Adults | Pharmaco-resistant or localization-related epilepsy | 70% in select patients sz freedom (3)       | Cognitive effects, surgery-related risks          |
| <b>VNS Therapy</b>      | 12 and older       | Pharmaco-resistant epilepsy; partial seizures       | 43% of pts >50% sz reduction at 3 years (4) | Voice alteration, cough, pharyngitis, dyspnea     |

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## Diagnosis?

- \*Observations from family, witnesses (detailed descriptions)
- Medical antecedents, obstetrical history & childhood development
- Physical & neurological exam
- Blood & urine tests
- Tests : EEG, CT scan, MRI, X-ray, PET scan, angiogram, SPECT
- Neurological evaluations, neuro-psychology, psychiatric evaluations, neuro-ophthalmology

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## Sz - Meds

- Common AEDs
- Common side effects
- Drug interactions
- Therapeutic levels
- Toxic levels

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## Treatment

- KETOGENIC DIET (produces ketones in urine)
- MEDICATION (antiepileptic medication - AED)
- VAGUS NERVE STIMULATOR
- SURGERY (Craniotomy)

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## Classic & Newer AEDS

**Newer AEDs**

- Primidone (Mysoline®) -> PB
- Clobazam (Frisium®) -> benzo
- Nitrazepam (Mogodon®) -> benzo
- Carbamazepine (Tegretol®) (**CBZ**)
- Divalproex (DVA)/Valproate/Valproic Acid (Epival®/Depakene®) **VPA** >GI SE
- Levetiracetam (Keppra®)
- Felbamate (Felbatol®) D/C d/t liver probs
- Vigabatrin (Sabril®) **Restricted** d/t vision probs
- Oxcarbazepine (Trileptal®) ->CBZ
- Gabapentin (Neurontin®) -> gaba
- Lamotrigine (Lamictal®) ->no P450!
- Topiramate (Topamax®)
- Pregabalin (Lyrica®) ->gaba

**N/A in Canada yet:**

- Tiagabine (Gabitril®)
- Zonisamide (Zonegran®)
- Rufinamide (Banzel®) (used for LGS)
- Lacosamide (Vimpat®)

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## Drug Levels

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|   |  |
|---|--|
| <ul style="list-style-type: none"> <li>• Carbamazepine (CBZ)<br/>17-54 µmol/L<br/>4-12 mcg/ml</li> </ul>  | <ul style="list-style-type: none"> <li>• Phenytoin (PHT)<br/>40-80 µmol/L<br/>10-20 mcg/ml</li> </ul>        |
| <ul style="list-style-type: none"> <li>• Phenobarbital (PB)<br/>65-150 µmol/L<br/>20-40 mcg/ml</li> </ul> | <ul style="list-style-type: none"> <li>• Valproic acid (VPA)<br/>350-800 µmol/L<br/>50-115 mcg/ml</li> </ul> |

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| Medication   | Systemic/Physical Effects   | CNS Effects   |
|--|---|---|
| Clonazepam (Rivotril)                                      | Drooling<br><br><i>Rare:</i><br>Rash<br>Paradoxical anger<br>Thrombocytopenia<br>Depression   | Sedation, dizziness<br>Risk of aspiration<br><br>Paradoxical reaction: disinhibition<br>↓ concentration<br>Anterograde amnesia<br><br>Ataxia<br>Nystagmus |
| Carbamazepine (Tegretol)<br><br>*CR tab < GI & CNS effects | Pruritic rash<br>↓ WBC, ↓ Vit D<br><br><i>Rare:</i><br>Aplastic anemia,<br>↑ LFTs (GGT/ALK),<br>Hyponatremia (SIADH)<br>Cardiac abnormalities<br>↓ T3/T4/Vit K<br>Alopecia, ocular effects,<br>Osteomalacia | N & V<br>Diplopia<br>Ataxia<br>Sedation, dizziness<br>Dyskinesia<br>Nystagmus   |

## Comparison of AEDs

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• Please see Word document

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| Medication   | Systemic/Physical Effects  | CNS Effects   |
|--|--|---|
| Valproic Acid (Depakene)<br><br>(VPA > GI SE)<br><br>Divalproex (Epival) | Alopecia<br>Abdominal cramps<br>Hyperammonemia<br>Menstrual disturbances<br><i>Rare:</i> ↓ platelet & WBC<br>Hepatotoxicity<br>Pancreatitis<br>Low carnitine<br>CAUTION: PCOS<br>Obesity (more common in ♀)<br>*SJS w/ Lamotrigine | Sedation, fatigue<br>Dizziness, ataxia<br>N & V<br>Confusion<br>Headache<br>Tremor                  |
| Gabapentin (Neurontin)   | Edema<br>Weight gain<br>Rash<br>Behavior Δ, irritability (kids)<br>↓ WBC<br>Low platelets (rare)<br>ECG changes (rare)   | Lethargy, fatigue<br>Dizziness, ataxia<br>Headache<br>N & V<br>Diplopia<br>Tremor<br>Slurred speech |

| Medication              | Systemic/Physical Effects   | CNS Effects  |
|-------------------------|---|--|
| Phenobarbital           | Rash<br>Sleep problems<br>↓ Vit D & K<br><i>Rare:</i> blood dyscrasias, liver toxicity  | Sedation, ataxia, dizziness<br>Nystagmus<br>↓ concentration & cognition<br>Behavior Δ, irritability (kids)           |
| Phenytoin (Dilantin)    | Hirsutism<br>Acne<br>Gingival hyperplasia (50%)<br>↓ folate/T4/Vitamin D & K levels<br>Rash<br>Osteomalacia<br>↑ LFTs<br>Blood dyscrasias | Ataxia, dizziness<br>Nystagmus<br>↓ concentration<br>Sedation<br>Dyskinesia, tremor<br>Arrhythmia<br>N & V, diarrhea |
| Ethosuximide (Zarontin) | Anorexia<br><i>Rare:</i> Rash (SJS), blood dyscrasias, behavioral Δ (kids)  | Drowsiness, dizziness<br>Hiccups<br>Headache<br>N & V, diarrhea  |

| Medication             | Systemic/Physical Effects   | CNS Effects   |
|------------------------|---|---|
| Lamotrigine (Lamictal) | **Rash (1 <sup>st</sup> month: gen. red morbilliform)<br>Abdominal pain<br>Alopecia<br><br><i>Rare:</i><br>SJS & toxic epidermal necrolysis<br>Hepatotoxicity<br>Tics in kids | Dizziness, Ataxia<br>N & V<br>Asthenia<br>Headache<br>Lethargy, fatigue<br>Blurred vision, diplopia   |
| Topiramate (Topamax)   | Diarrhea<br>Weight loss<br>Kidney stones<br>Glaucoma<br>Rare: ↑ LFTs  | Drowsiness, fatigue<br>Headache<br>Dizziness, ataxia<br>Agitation<br>Behavioral Δ<br><br>Paresthesias (fingers, toes)<br>Cognitive deficits (memory, concentration, word-finding) |

## VNS & Sx for Sz control

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- Indications?
- Contraindications?
- Common risks?
- Outcomes/benefits?
- Post-op complications?

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## SOLUTION-S

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Ottawa, Ontario K1B 5H3  
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info@solution-s.ca  
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## Useful Tools!

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- Scatterplot/calendar
- Observation sheets (epilepsy)
- VIDEO of the 'episodes'

\*\*important to document all changes in medication, especially if there have been any recent changes

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## Key aspects to document:

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- Activity before the seizure?
- Activity during the seizure? Note sequence of events exactly: movements of eyes, head, arms & legs. Does the person respond to you during the episode?
- How did they behave after the seizure?
- Is there a pattern?
- When does it happen, during favorite activities or only those which are disliked?

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**Comparison of AEDs**

|                 | Carbamazepine (CBZ)   | Oxcarbazepine (OXC)  | Valproic Acid/Divalproex (VPA/DVA)   | Gabapentin (GBP)                                     | Topiramate (TPM)  |
|-----------------|---|--|--|--|---|
| Doses           | 600-1800mg/day<br>BID-TID dosing<br><br>*available in CR form   | 600-1200mg/day<br>in divided doses   | 750-3000mg/day<br>BID-TID dosing   | 900-3600mg/day<br>TID dosing                         | 200-600mg/day<br>BID dosing   |
| Meta-<br>bolism | Liver & P-gp<br>* induces own<br>metabolism   | Liver<br>* DOES NOT induce<br>own metabolism   | Liver  | Not metabolized,<br>Eliminated by<br>renal excretion | P-gp, ( 70% is<br>eliminated<br>unchanged in urine)   |
| Drug<br>levels  | 17-54 µmol/L<br>4-12 mcg/ml<br>* 5d after Rx started & 5d after Δ<br>dose or +/- other Rx                 | Not required   | 350-800 µmol/L<br>50-115 mcg/ml<br>* 5d after Rx started & 5d after Δ dose<br>or +/- other Rx  | Not required   | Not required  |
| W/U             | 1. CBC, plats & diff<br>2. E-<br>3. LFTs<br>4. TSH<br>5. ECG (>45yrs)<br>6. BMD                           | E-   | 1. CBC, plats & diff<br>2. LFTs<br>3. Lipid profile<br>(total, HDL & TG)<br>4. ♀: wt & BMI<br>5. Consider serum<br>testosterone in young ♀<br>6. BMD   | serum Cr (sCr)                                       | Baseline serum<br>bicarbonate<br>sCr  |
| F/U             | Repeat #1 after 1 <sup>st</sup> month,<br>then 2-3X/yr.<br>Repeat #2 q.6months.<br>Repeat #2 - 4 annually | CBC, plats, E-, TSH, LFTs<br>annually.<br>Na+ levels when<br>suspected hyponatremia. | Repeat #1 & 2 monthly X2, then<br>2-3X/yr.<br>Repeat #3 & 4 annually.<br>Test #5 if s/s of menstrual<br>hyperandrogenism.<br>Test prolactin, LH & TSH for<br>insulin resistance & HTN.<br>Ammonia levels if lethargy & Δ<br>LOC. | LH & TSH<br>sCr if renal toxicity<br>suspected       | Periodic serum<br>bicarbonate;<br>sCr if renal toxicity<br>suspected<br>(risk of kidney stones) |

**Comparison of AEDs**

|                 | Lamotrigine (LTG)           | Levetiracetam (LEV)   | Zonisamide (ZNS) (*sulfa Rx)                                 | Tiagabine (TGB)               | Phenobarbital (PB)                          | Phenytoin (PHT)                                     |
|-----------------|-----------------------------|---|--|-------------------------------|---|---|
| Doses           | 100-500mg/day<br>BID dosing | 1000-3000<br>mg/day<br>BID dosing   | 100-600mg/day<br>in single or<br>BID dosing                  | 32-56mg/day<br>BID-QID dosing | 15-180mg/day in single<br>or divided doses  | 300-400mg/day in<br>single or divided doses         |
| Meta-<br>bolism | Liver                       | Not metabolized,<br>Eliminated by<br>renal excretion<br>( 66% eliminated<br>unchanged in urine) | Liver  | Liver                         | Liver                                       | Liver   |
| Drug<br>levels  | Not required                | Not required  | Not required   | Not required                  | 65-150 µmol/L<br>20-40 mcg/ml               | 40-80 µmol/L<br>10-20 mcg/ml                        |
| W/U             | CBC, LFTs                   | CBC, plats & diff,<br>sCr   | CBC & diff, LFTs, sCr  |                               | CBC & diff, LFTs                            | CBC & diff, LFTs,<br>folate?                        |
| F/U             | CBC, LFTs<br>annually       | CBC & diff, sCr<br>annually   | CBC & diff, LFTs, sCr<br>annually (risk of kidney<br>stones) | none                          | CBC & diff, LFTs<br>annually.<br>BMD/Vit D* | CBC & diff, LFTs, folate<br>annually.<br>BMD/Vit D* |

Virani, A., Bezchlibnyk-Butler, K., & Jeffries, J., *Clinical Handbook of Psychotropic Drugs*, (2009); *Rx Files*, (2008).

**\*Use alphabetic order to recall drug levels (in mcg/ml units):** CBZ (4-12), Dilantin (10-20), PB (20-40), VPA (50-115)

## **GENETIC SYNDROME WEBSITES**

### **Angelman Syndrome**

Angelman Syndrome Foundation, Inc. (USA) Website : [www.angelman.org](http://www.angelman.org)  
Canadian Angelman Syndrome Society Website : [www.angelmancanada.org](http://www.angelmancanada.org)

### **Cornelia de Lange Syndrome**

Cornelia de Lange Syndrome Foundation, Inc. (USA) Website : [www.cdlsusa.org](http://www.cdlsusa.org)  
Cornelia de Lange Syndrome, Canada Website : [www.cdlsCanada.ca](http://www.cdlsCanada.ca)

### **Cri-du-Chat Syndrome**

UK Cri Du Chat Syndrome Support Group (UK) Website : [www.criduchat.co.uk](http://www.criduchat.co.uk)  
5p minus Society (USA) Website: <http://www.fivepminus.org/>

### **Down Syndrome**

Down Syndrome Research Foundation (Canada) Website: [www.dsrf.org](http://www.dsrf.org)  
Canadian Down Syndrome Society Website: [www.cdss.ca/](http://www.cdss.ca/)  
Down Syndrome Association of Ontario Website: [www.dsao.ca/](http://www.dsao.ca/)  
Down Syndrome Association - National Capital Region Website: [www.dsancr.com/](http://www.dsancr.com/)  
National Down Syndrome Society (USA) Website: <http://www.ndss.org/>  
National Association for Down Syndrome (USA) Website: [www.nads.org](http://www.nads.org)  
National Down Syndrome Congress Website: [www.ndsccenter.org](http://www.ndsccenter.org)  
Down Syndrome Association-UK Website: [www.dsa-uk.com](http://www.dsa-uk.com)  
Down Syndrome Health Issues, Dr Len Leshin website: [www.ds-health.com](http://www.ds-health.com)  
International Mosaic Down Syndrome Association website: [www.imdsa.org](http://www.imdsa.org)

### **Fragile X Syndrome**

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

### **Neurofibromatosis**

Neurofibromatosis, Inc. Website: [www.nfinc.org](http://www.nfinc.org)  
Children's Tumor Foundation Website: [www.ctf.org](http://www.ctf.org)

### **PKU**

National PKU News Website: <http://www.pkunews.org/>

### **Prader-Willi Syndrome**

The Prader-Willi Syndrome Association (PWSA) (USA) Website: [www.pwsausa.org](http://www.pwsausa.org)  
Prader-Willi Alliance of NY. (USA) Website: [www.prader-willi.org](http://www.prader-willi.org)

### **Rett Syndrome**

International Rett Syndrome Association (USA) Website: [www.rettsyndrome.org](http://www.rettsyndrome.org)  
Rett Syndrome Association UK Website: [www.rettsyndrome.org.uk](http://www.rettsyndrome.org.uk)

## **GENETIC SYNDROME WEBSITES...**

### **Smith-Lemli-Opitz Syndrome**

Smith-Lemli-Opitz/RSH Foundation Website: [www.smithlempiopitz.org](http://www.smithlempiopitz.org)

### **Smith-Magenis Syndrome**

Parents and Researchers interested in Smith-Magenis Syndrome (PRISMS) (USA)

Website: [www.smithmagenis.org](http://www.smithmagenis.org)

### **Tuberous Sclerosis**

Tuberous Sclerosis Alliance (USA) Website: [www.tsalliance.org](http://www.tsalliance.org)

### **Williams Syndrome**

Williams Syndrome Foundation (USA) Website: [www.wsf.org](http://www.wsf.org)

The William Syndrome Association (WSA) (USA) Website: [www.williams-syndrome.org](http://www.williams-syndrome.org)

### **22q11 Deletion (DiGeorge/Velocardiofacial/Shprintzen) Syndrome**

22q-11/Velo-Cardio-Facial Syndrome (VCFS)/Di George Syndrome/The Shprintzen Syndrome

VCFS Educational Foundation (USA) Website: [www.vcfsef.org](http://www.vcfsef.org)

22q and You Newsletter (USA) Website: [www.cbil.upenn.edu/VCFS/22qandyou/](http://www.cbil.upenn.edu/VCFS/22qandyou/)

VCFS International Center (Upstate University Hospital) [www.upstate.edu/ent/velo.shtml](http://www.upstate.edu/ent/velo.shtml)

### **Other Genetics 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](http://www.surreyplace.on.ca/Clinical-Programs/Medical-Services/Pages/PrimaryCare.aspx)

Geneclinics : <http://geneclinics.org/> (Info under: 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 Education Center, University of Kansas Medical Center:

<http://www.kumc.edu/gec/support/>

The Family Village: <http://www.familyvillage.wisc.edu>

### **Genetics Website (In English, French, Spanish, German, Italian & Portuguese!):**

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