



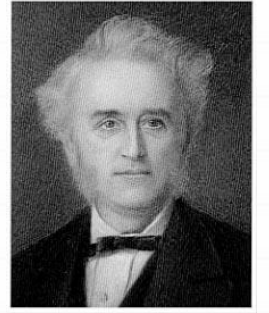
UCSF Benioff Children's Hospital
Oakland

Comprehensive
care of children
with
Trisomy 21

I'm "up" on Down
syndrome

Disclosures

- 0 No one involved in the planning or presentation of this activity has any relevant financial relationships with a commercial interest to disclose.
- 0 I really love talking about this, I may get a bit animated



What is Down Syndrome?

- 0 An extra copy of the 21st chromosome
 - 0 Meiotic non-disjunction (96%)
 - 0 Translocation (3-4%)
 - 0 Mosaicism (1-2%)
- 0 One in 800 live births
 - 0 Over 200,000 affected in US
- 0 Each child is different in their phenotype
 - 0 Many associated conditions
 - 0 Quite anxiety provoking, especially for new parents

Diagnosis: Prenatal

- 0 Prenatal detection becoming more common – often incidental to sex determination
 - 0 Cell Free DNA is a screening test
 - 0 Cell Free DNA is not diagnostic - 99.7% detection rate and 0.04% false positive rate
 - 0 Diagnostic testing = Amniocentesis or Chorionic Villus sampling
- 0 Allows prenatal diagnosis of co-occurring conditions
- 0 Opportunity for optimal delivery arrangement
- 0 Opportunity to offer parent-to-parent contact and support resources
- 0 Can be associated with trauma if news are delivered poorly

Prenatal Diagnosis Support

- 0 Jack's Basket
- 0 Down Syndrome Diagnosis Network
- 0 Local support organizations:
 - 0 Down Syndrome connection of the Bay Area
 - 0 Silicon Valley Down Syndrome Network

Just Released: *The Prenatal Consult Film*



This new resource is designed for healthcare providers to share with expectant parents following a prenatal diagnosis of Down syndrome. This short 17-minute film features perspectives from both providers and parents of children with Down syndrome with the goal to:

- shed light on some of the 'unknowns' that families may face when receiving a prenatal Down syndrome diagnosis,
- encourage expectant parents with stories and experiences from other families who are raising a child with Down syndrome,
- and to most importantly offer hope for their journey ahead.

This short film was created in partnership with the Jack's Basket medical team and features three providers and four families, each sharing their own unique experiences caring for babies with Down syndrome and raising them to thrive.

[WATCH NOW](#)



Expectant Parent Resources

An unexpected confirmed or likely Down syndrome diagnosis and pregnancy can be a challenging time for parents. We know how important it is to find resources to meet your needs, so we have compiled several resources to help you navigate this time.

Online Support

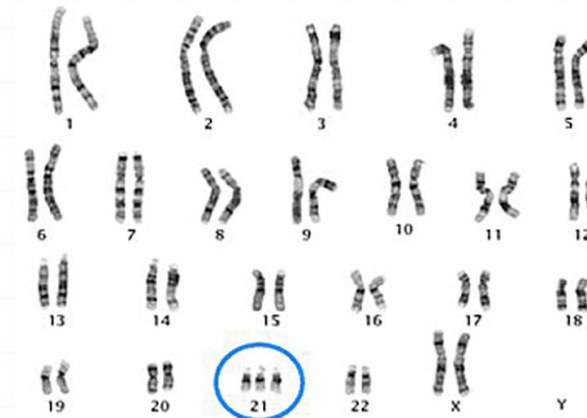
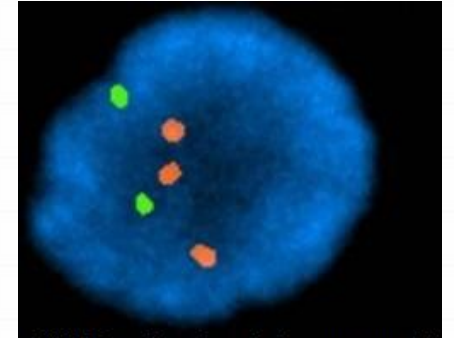
(for those continuing a pregnancy)

- [DSDN Pregnancy Group](#)



Diagnosis: Post-natal

- 0 Confirmatory tests:
 - 0 FISH = fast (24-48 hours)
 - 0 Karyotype = slow
 - 0 Tells translocations from non-disjunction
 - 0 **Required** for diagnosis after delivery
 - 0 Critical for genetic counseling
 - 0 Chromosomal Microarray Analysis (CMA) = slow
 - 0 Can tell about other genetic differences as well
 - 0 Still not enough for genetic counseling



What Should I Say?

Pediatrics
October 2009, VOLUME 124 / ISSUE 4

Postnatal Diagnosis of Down Syndrome: Synthesis of the Evidence on How Best to Deliver the News

Brian G. Skotko, George T. Capone, Priya S. Kishnani



What Should I Say?

- 0 Please start with CONGRATULATIONS. Refer to the baby by their name. Use person-first language
- 0 “I would like to discuss some news you **may not have been expecting**”
- 0 “Is this a good time to talk? Before we start talking, **should anyone else be here with you?**”
- 0 You don’t need to wait until you have a confirmatory diagnosis
- 0 Discuss relevant findings/clinical manifestations
 - 0 Start listing off all associated conditions
 - 0 Start listing off all the referrals that will be needed
- 0 Refer to genetic counseling

**HOW TO DELIVER A
DOWN SYNDROME
DIAGNOSIS.
THE RIGHT WAY.**

Welcome to Holland

by Emily Perl Kingsley

When you're going to have a baby, it's like planning a fabulous vacation trip - to Italy. You buy a bunch of guide books and make your wonderful plans. The Coliseum. The Michelangelo David. The gondolas in Venice. You may learn some handy phrases in Italian. It's all very exciting.

After months of eager anticipation, the day finally arrives. You pack your bags and off you go. Several hours later, the plane lands. The stewardess comes in and says, "Welcome to Holland."

"Holland?!?" you say. "What do you mean Holland?? I signed up for Italy! I'm supposed to be in Italy. All my life I've dreamed of going to Italy."

But there's been a change in the flight plan. They've landed in Holland and there you must stay.

The important thing is that they haven't taken you to a horrible, disgusting, filthy place, full of pestilence, famine and disease. It's just a different place.

So you must go out and buy new guide books. And you must learn a whole new language. And you will meet a whole new group of people you would never have met.

It's just a different place. It's slower-paced than Italy, less flashy than Italy. But after you've been there for a while and you catch your breath, you look around.... and you begin to notice that Holland has windmills.... and Holland has tulips. Holland even has Rembrandts.

But everyone you know is busy coming and going from Italy... And for the rest of your life, you will say "Yes, that's where I was supposed to go. That's what I had planned."

But... if you spend your life mourning the fact that you didn't get to Italy, you may never be free to enjoy the very special, the very lovely things... about Holland.

Health Maintenance

- 0 They are a child like any other, so we do the same screenings for health supervision
- 0 What we know about DS informs additional screenings

American Academy
of Pediatrics



DEDICATED TO THE HEALTH OF ALL CHILDREN™

Health Supervision for Children and Adolescents With Down Syndrome

Marilyn J. Bull, MD, FAAP,^a Tracy Trotter, MD, FAAP,^b Stephanie L. Santoro, MD, FAAP,^c Celanie Christensen, MD, MS, FAAP,^a
Randall W. Grout, MD, MS, FAAP,^d THE COUNCIL ON GENETICS

Neonatal Period (birth-1mo)

Neonatal Period

What needs to be addressed right away?

- 0 **Eyes** – cataracts
- 0 **Ears** – newborn hearing screen to look for congenital hearing loss
- 0 **Thyroid** – congenital hypothyroidism. Send thyroid studies
- 0 **Respiratory** – evaluate for airway issues clinically
 - 0 **Evaluate for A/B/D** – Car seat test
- 0 **Heart** –congenital heart defects (50%)
 - 0 Must get an echo even if fetal echo was normal
 - 0 Increased chance of pulmonary hypertension, even in the absence of CHD
- 0 **Blood** – CBC to look for transient abnormal myelopoiesis (10%), polycythemia
 - 0 TAM can be symptomatic (jaundice, rash, big liver/spleen) or asymptomatic
 - 0 History of TAM increases lifetime risk for leukemia

Neonatal Period

What needs to be addressed right away?

Feeding

- 0 Feeding difficulties present in 30-80% of infants with DS
 - 0 Infants with Down syndrome can breastfeed successfully, but some may need early support until a successful nursing pattern is established
 - 0 90% of infants with DS who aspirate do so silently
 - 0 Refer for feeding evaluation and possible swallow study if:
 - 0 Marked hypotonia as judged by the pediatrician
 - 0 Underweight
 - 0 Slow feeds
 - 0 Choking with feeds
 - 0 Recurrent or persistent respiratory symptoms
 - 0 Desaturations with feeds

Gut

- 0 Structural GI defects
- 0 Hirschsprung's disease

Neonatal Period: Anticipatory Guidance

- 0 **Strengths, potential**
- 0 Early intervention services
- 0 Protecting from resp. infections
 - 0 Flu vax for the whole family, TDaP booster
 - 0 Synagis?
- 0 Local support groups, how to talk to sibs
- 0 Support the neck, car seat

Infancy (1mo-1yr)

Focus on growth, development, staying healthy

Infancy (1mo to 1 yr)

- 0 **Growth** – plot on both the WHO and DS specific growth charts
- 0 **Feeding and gut** – Screen for dysphagia. Screen for and support reflux. Screen for and support constipation
- 0 **Eyes** –Refer within the first 6 months.
 - 0 May see nystagmus, cataracts, strabismus, lacrimal duct stenosis
- 0 **Ears** – Even if they passed newborn hearing screening, audiology eval at 6 mo, 1 yr (more often if documented hearing loss, if child is prescribed a hearing aid)
 - 0 ENT if any new concerns
 - 0 In children with stenotic canals in which the tympanic membranes cannot be seen, refer to ENT for ear exam with office microscope
- 0 **Heart** – CHD repaired or monitored. Watch for CHF, poor growth

Infancy (1mo to 1 yr)

- 0 **Thyroid** – Review newborn labs. Repeat at 6 and 12 months. Repeat sooner if the child shows symptoms of hypothyroidism
- 0 **Neuro** – Increased risk of infantile spasms. Teach families signs of myelopathy
- 0 **Blood** – Screen for anemia with a CBC + iron studies or CBC + ferritin and CRP.
 - 0 MCV often high, cannot use it to screen for iron deficiency
 - 0 Increased risk of leukemia
- 0 **Resp** – Airway malacia is common. URIs can be more difficult to manage. Prevention is key with immunizations and infection prevention measures
- 0 **Dental** – irregular and delayed eruption is common. Oral desensitization is helpful

Developmental Monitoring

- 0 Children with Down syndrome typically will have some degree of developmental delay
- 0 It is important to ask open ended questions about developmental progress at each visit
 - 0 When a parent says “no”, I rephrase it as “not yet”
- 0 You are watching for developmental plateaus and regression
- 0 Ensure connection to early intervention for developmental support

Infancy: Anticipatory Guidance

- 0 How is the family doing? (don't forget the sibs)
- 0 Weight, growth curves
- 0 Early intervention services, RCEB
- 0 Again share information about support organizations
- 0 Support the neck, car seat
- 0 Ensure the family has had access to genetic counseling
- 0 Alternative/complementary medicine

Early Childhood (1-5 yrs)

Early Childhood (1-5 yrs)

- 0 Goal: enable the child to achieve their maximal potential
- 0 Monitor and address anything that affects learning
 - 0 Hearing
 - 0 Vision
 - 0 OSA
 - 0 Thyroid
 - 0 Discomfort
 - 0 Autism

Early Childhood (1-5 yrs)

- 0 **Eyes** – yearly checks. 50% will have a refractive error and need glasses
 - 0 If untreated, can lead to vision loss
- 0 **Ears** – Checks with audiology every 6 months if there is hearing loss or until ear-specific information can be obtained, otherwise yearly
 - 0 50-70% have serous otitis
- 0 **Feeding and gut**
 - 0 Review feeding patterns
 - 0 Chewing difficulties? Sensory preferences? At risk for nutritional deficiencies
 - 0 Review growth (BMI on CDC curves) and watch out for weight gain.
 - 0 Review symptoms of GERD, constipation

Early Childhood (1-5 yrs)

- 0 **Heart** – Ensure adequate follow up with cardiology. Give PPSV23 when indicated
- 0 **Endocrine** – Check thyroid at least annually.
 - 0 Watch for symptoms of celiac disease and screen
 - 0 Watch for signs of diabetes
- 0 **Blood** – Review intake of iron rich foods. Screen for anemia with yearly CBC + iron studies. Watch for signs of leukemia
 - 0 Same or lower risk for most solid tumors EXCEPT testicular tumors
 - 0 Must check the testicles at every check up, review privacy concepts

Early Childhood (1-5 yrs)

- 0 **Neuro** – discuss cervical spine positioning signs and symptoms of atlantoaxial instability
 - 0 Change in gait or use of arms or hands
 - 0 Change in bowel or bladder function
 - 0 Neck pain
 - 0 Stiff neck
 - 0 Head tilt, torticollis, how the child positions his or her head
 - 0 Change in general function
 - 0 New weakness.
- 0 Screening xrays are not helpful. However, if a child is symptomatic, they will need imaging and neurosurgery evaluation
- 0 Avoid activities that cause whiplash and hyperextension
 - 0 Careful with dentist, anesthesia, office exam

Early Childhood (1-5 yrs)

- 0 **Obstructive Sleep Apnea** (30-90% of children)
 - 0 **9 point lower IQ scores**
 - 0 Linked with behavior problems, ADHD
 - 0 Can have negative impacts on the heart and cardiovascular system
 - 0 Increased risk of abnormal glucose levels and diabetes
- 0 Symptom based questionnaires underestimate prevalence
- 0 May see: snoring with pauses, abnormal sleep positions (neck hyper-extended, sitting up bent at the hip), frequent awakenings, daytime fatigue/difficulty waking up
- 0 Multilevel problem
 - 0 Tonsils+ adenoids
 - 0 Tongue
 - 0 Airway wall collapse
- 0 Size of tonsils not necessarily predictive of OSA
- 0 **Bottom line: screen everyone (LATEST is 4 yo)**

Early Childhood (1-5 yrs) Behavioral Support

0 Difficult behaviors can be commonly seen

0 The 'flop'

0 Throwing/flinging

0 Dangling

0 Eloping

0 Contributing factors:

0 Difficulty communicating

0 Difficulty with transitions

0 Crowds can be overwhelming

0 Sensory integration issues

Early Childhood (1-5 yrs)

Behavioral Support

What is the purpose of the behavior?

Escape from, delay, or avoid, tasks and activities

0 Ex: demands, chores, transitions

Gain Attention from adults or other students

0 Ex: parent/teacher attention, peer attention, reprimands, praise

Access Tangible/preferred items (Toys/Things)

0 Ex: Preferred activities, toys, books, snacks, etc.

Sensory

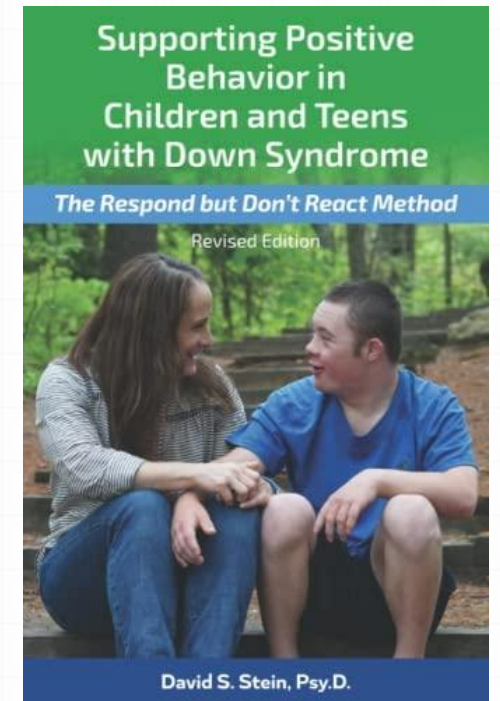
0 The behavior may “feel good” or be calming to the student

0 Ex: visual stimulation, auditory stimulation, rocking, tapping, etc.



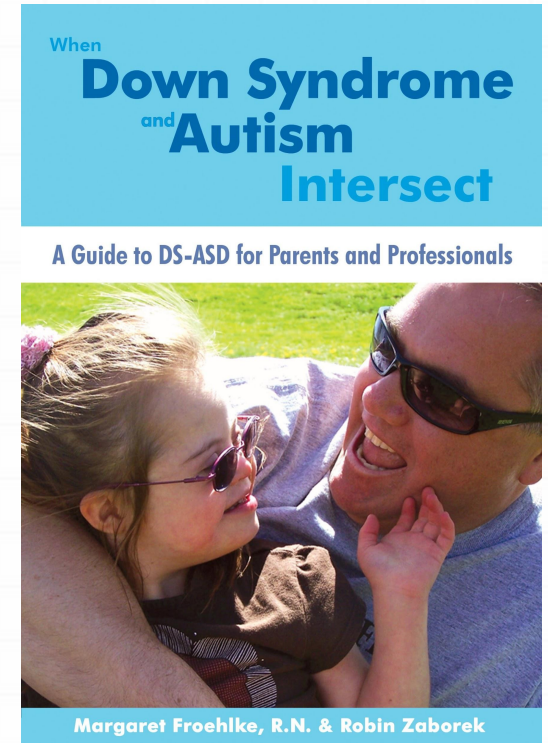
Early Childhood (1-5 yrs) Behavioral Support

- 0 Evaluate for medical problems that can be associated with behavior changes
 - 0 Thyroid abnormalities, celiac disease, sleep apnea, gut discomfort
- 0 Support positive behavior
 - 0 Control antecedents whenever possible
 - 0 Respond, don't react
 - 0 Reward desired behavior (avoid food rewards)
- 0 May need referrals:
 - 0 Developmental evaluation (ADHD, Autism, etc)
 - 0 Psychosocial services
 - 0 Behavioral specialists
- 0 Careful use of medication for behavior management
 - 0 Children with Down syndrome may be more sensitive to psychotropic medication



DS+ Autism Spectrum

- 0 At least 10% of kids with Down syndrome also have autism
 - 0 Best estimate: 16-18%
- 0 **Challenging diagnosis**
 - 0 Diagnostic overshadowing
 - 0 Developmental delays can confound the picture
 - 0 Medical issues can delay the diagnosis
 - 0 Language delays are present in most individuals with DS
- 0 Recognition is imperative – leads to appropriate and timely intervention



DS-Autism Profile

- 0 Lack of joint attention
- 0 Self directed, lack of engaging in reciprocal play
 - 0 Play may be repetitive, prefer cause/effect toys
- 0 Lack of non-verbal communication
 - 0 Gestures, pointing
- 0 Less interested in overall communication
 - 0 May still be able to repeat words/signs
- 0 Texture sensitivity
 - 0 Food, clothing/diaper, sand/water/grass

DS-Autism Profile

- 0 Hypo/hyperactive
- 0 Gaze aversion, inconsistent eye contact
- 0 Regression of plateau of skills
- 0 Motor mannerisms
 - 0 Many kids with DS will have self-stimulatory behaviors
 - 0 DS-ASD are harder to redirect
 - 0 Would rather stim than engage with you
- 0 Inflexible/rigid
- 0 Sociability often retained!
 - 0 May still smile, be more “loving”

DS+ Autism Spectrum

- 0 Screening tools:
 - 0 Can still use MCHAT
 - 0 Consider social communication questionnaires
- 0 Diagnostic
 - 0 ADI-R and ADOS can catch up to 70%
 - 0 Doesn't change who the child is, helps us explain and treat differences
- 0 Treatment:
 - 0 Must treat autism in order to maximize learning
 - 0 ABA
 - 0 Appropriate educational program
 - 0 Floortime, encourage reciprocal play, encourage imitation
 - 0 Augmentative communication modalities (but don't give up on speech!)
 - 0 Sensory support

Early Childhood: Anticipatory Guidance

- 0 Optimal nutrition, encourage physical activity
- 0 Immunizations and infection preventions
- 0 Developmental support, transition to school system:
 - 0 IFSP (Regional Center) □ IEP (school district)
- 0 Socialization opportunities
- 0 Encourage use of accurate terms for genitalia and other private body parts
 - 0 Discuss abuse prevention
- 0 Neck support: avoid trampolines etc, appropriate car restraints

Late Childhood (5-12 years)

Late Childhood (5-12 years)

- 0 **Eyes** – vision should be checked yearly, ophthalmologist every 2 years
- 0 **Ears** – hearing test yearly
- 0 **Feeding and gut** – growth, GERD, constipation
 - 0 Watch out for weight gain!
- 0 **Heart** – Does the child need prophylaxis at the dentist? Activity restrictions? Repeat surgeries?
- 0 **Endocrine** – Check thyroid annually. Get antibody titers if screening labs are abnormal.
 - 0 Ongoing risk of developing acquired hypothyroidism, celiac disease, type I and II diabetes
- 0 **Neuro** – screen for myelopathy, seizures
- 0 **Blood** – annual CBC
- 0 **Resp** – Continue to screen for and manage sleep apnea. Intervene as indicated
- 0 **Skin** – Complete skin exam (psoriasis, alopecia, vitiligo, fungal infections)

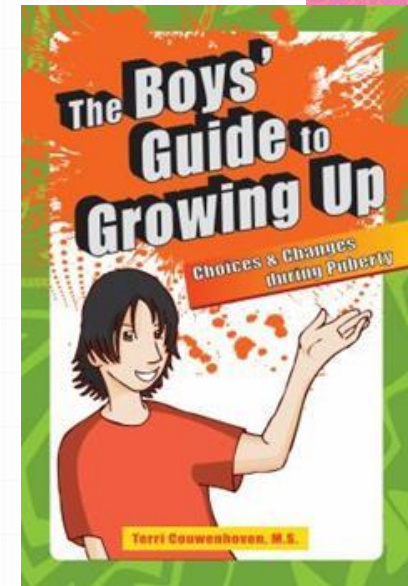
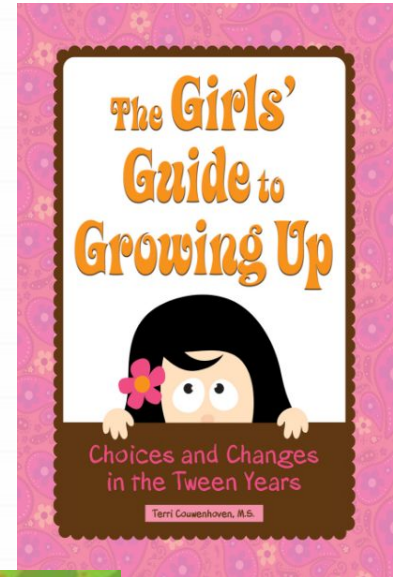
Late Childhood (5-12 years)

Promoting Self Help Skills

- 0 What could the child be doing alone?
 - 0 Encourage gradual fading of assistance where appropriate
- 0 What chores does the child have?
 - 0 Chores allow practicing of skills in a safe environment
 - 0 Can translate to future employment skills
- 0 How does the child's educational program support independence?
 - 0 Formal discussion of transition will begin later, but can have independence and self-advocacy goals throughout
- 0 When is it possible to do shared decision-making?

Anticipatory Guidance

- 0 Behavioral and psychological issues
 - 0 Screen and refer as appropriate
- 0 Encourage and support socialization
- 0 Transition from elementary to middle school
 - 0 From 1 to many teachers and from 1 class to changing classes.
- 0 Independence with hygiene and self care
- 0 Discuss puberty
 - 0 Teach, model, and respect privacy
 - 0 Discuss sexual health
 - 0 Safety and abuse prevention



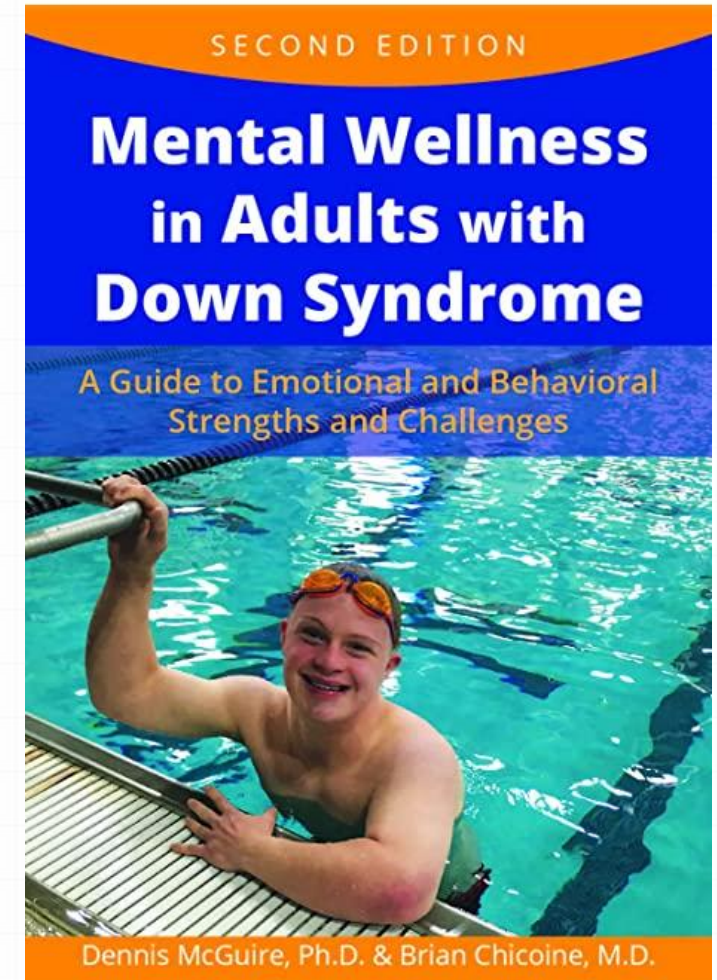
Adolescence to Early Adulthood (12-21 years)

Adolescent/Early Adulthood (12-21 years)

- 0 **Eyes** – vision should be checked yearly, ophthalmologist every 3 years
 - 0 Watch for keratoconus
- 0 **Ears** – hearing test yearly
 - 0 Watch for early onset presbycusis
- 0 **Feeding and gut** – growth, digestive issues
 - 0 Ongoing monitoring and management of obesity
- 0 **Heart** – Does the child need prophylaxis at the dentist? Activity restrictions? Repeat surgeries?
 - 0 Monitor for acquired mitral/aortic valvular disease
- 0 **Thyroid** – annually. Ongoing risk of developing acquired hypothyroidism, celiac disease, type I and II diabetes
- 0 **Neuro** – screen for myelopathy, seizures
- 0 **Blood** – annual CBC
- 0 **Resp** – Continue to screen for and manage sleep apnea. Intervene as indicated

Adolescent/Early Adulthood Mental Wellness

- 0 High rates of anxiety, depression, other mental health issues
- 0 Down syndrome regression
 - 0 Loss of skills, acute or subacute, with psychiatric and neuro-immunologic causes
 - 0 Too late for autism regression, too early for dementia
- 0 Difficult to find qualified providers
- 0 Social health an important protective factor



Anticipatory Guidance

- 0 Transition to adulthood
 - 0 Decision making
 - 0 Long term financial planning
 - 0 Vocational training as part of the IEP
 - 0 Living arrangement
- 0 Social network, connectedness
- 0 Sexual health
 - 0 Family planning
 - 0 Safety
- 0 Facilitate transition to adult care



The End!
Questions???

References

1. Marilyn J. Bull and the Committee on Genetics. Health Supervision for Children With Down Syndrome. *Pediatrics* 2011; 128; 393
2. Skotko BG, Capone GT, Kishnani PS; Down Syndrome Diagnosis Study Group. Postnatal diagnosis of Down syndrome: synthesis of the evidence on how best to deliver the news. *Pediatrics*. 2009 Oct;124(4):e751-8
3. Stein, David. Supporting Positive Behavior in Children and Teens with Down Syndrome. The Respond but Don't React Method. Bethesda, MD: Woodbine House, 2016
4. Levine MD, Carey WB, Crocker AC, editors: *Developmental-behavioral pediatrics*, ed 2, Philadelphia, 1992, Saunders.
5. Rasmussen P, Börjesson O, Wentz E, Gillberg C. Autistic disorders in Down syndrome: background factors and clinical correlates. *Dev Med Child Neurol*. 2001;43(11):750.
6. Zemel BS, Pipan M, Stallings VA, Hall W, Schadt K, Freedman DS, Thorpe P. Growth Charts for Children With Down Syndrome in the United States. *OPediatrics*. 2015 Nov;136(5):e1204-11.