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Pediatric Healthcare Guidelines

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Medical Outreach Director, DSANEO





American Academy of Pediatrics Health Supervision for Children and Adolescents with Down Syndrome

Guidelines were first created in 1994. They were revised in 2011, and most recently in 2022.

<https://publications.aap.org/pediatrics/article/doi/10.1542/peds.2022-057010/186778/Health-Supervision-for-Children-and-Adolescents>



Birth

At birth, the baby will get a thorough evaluation looking for any medical issues, especially:

a heart problem

a gastrointestinal problem

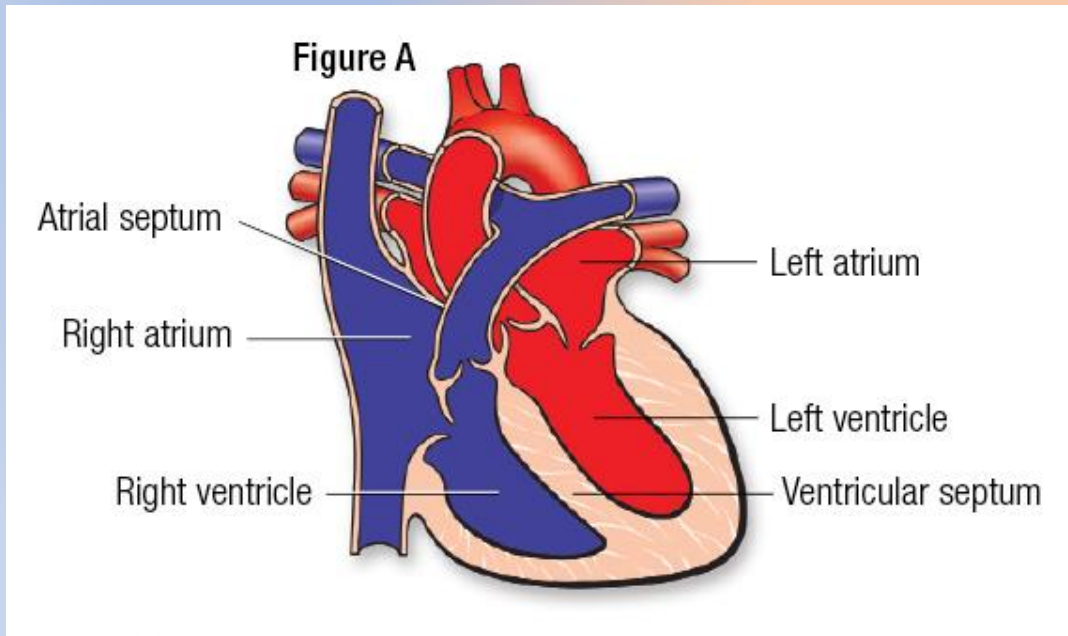
feeding issues

an eye problem, such as cataracts

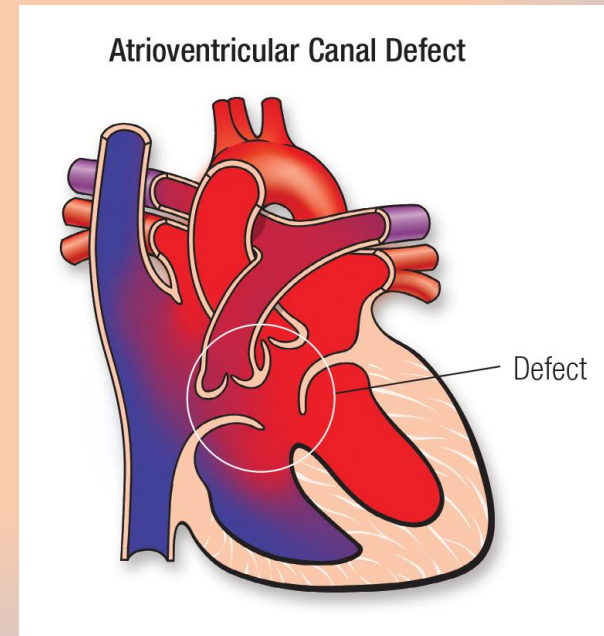
Heart Problems

<https://www.heart.org/en/health-topics/congenital-heart-defects>

Typical Heart Structure



AV Canal



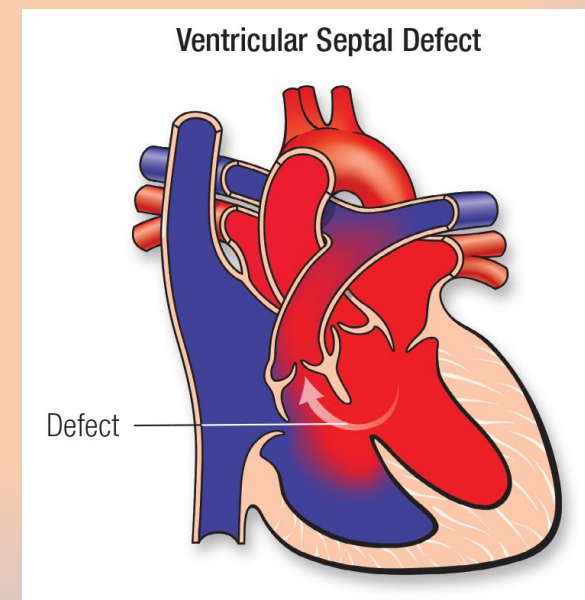
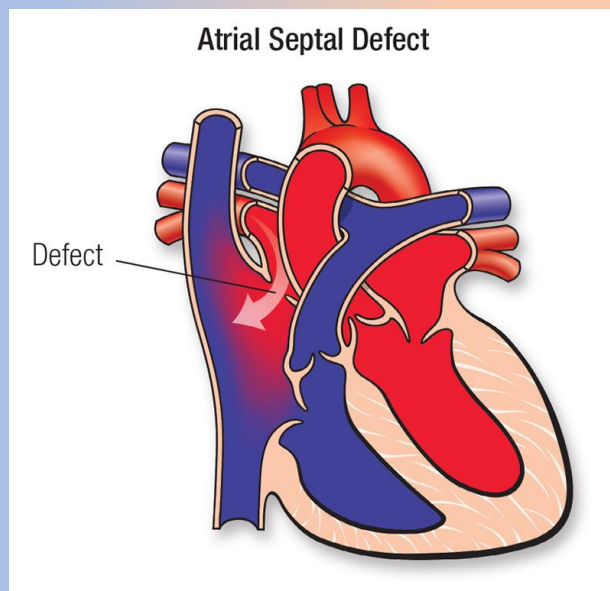
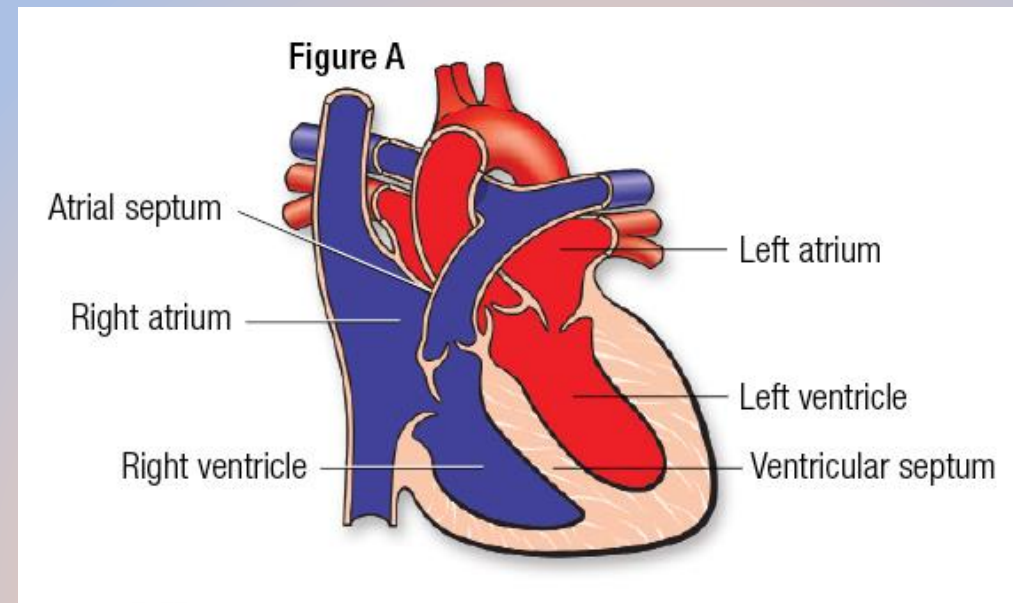
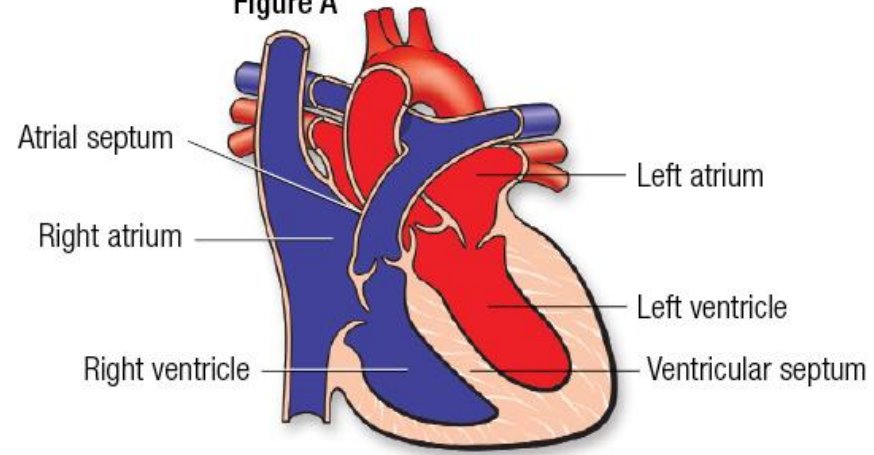
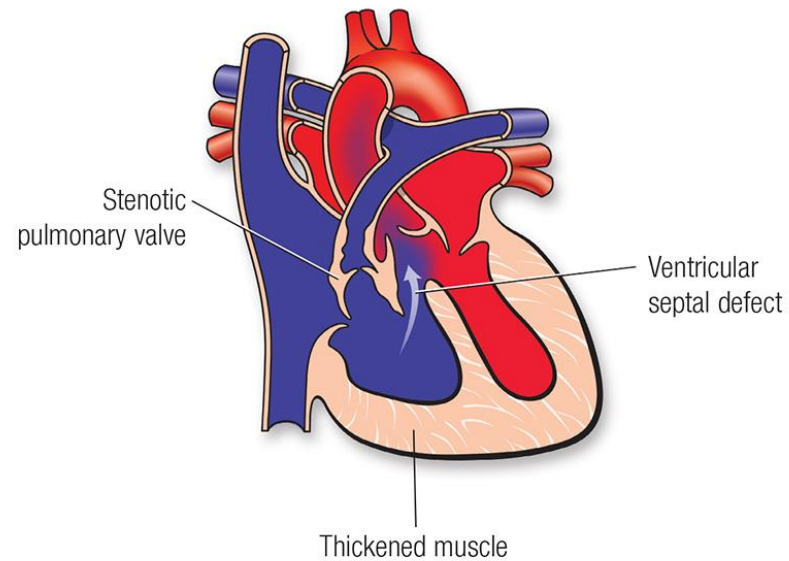


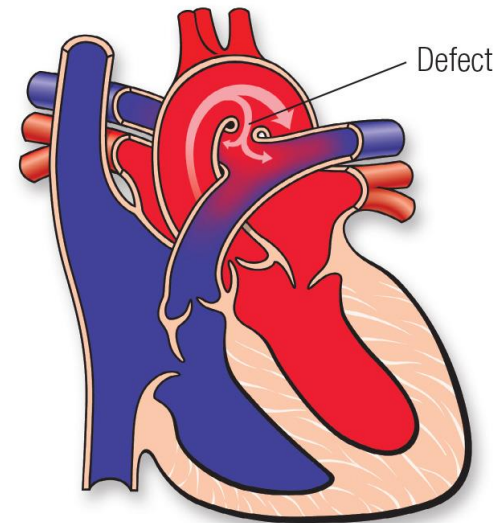
Figure A



Tetralogy of Fallot

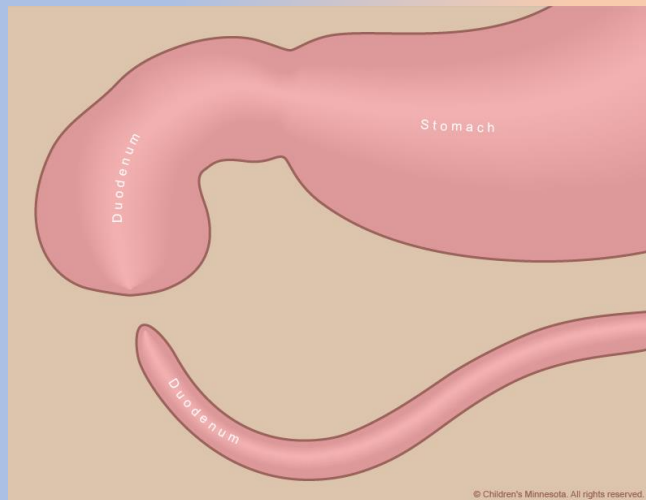


Patent Ductus Arteriosus

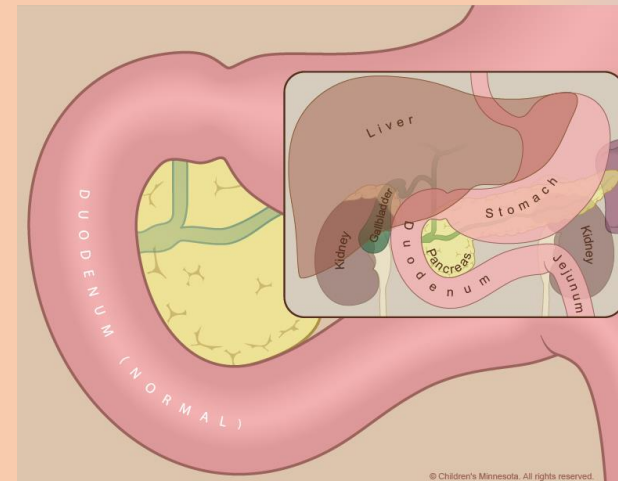


Gastrointestinal Problems

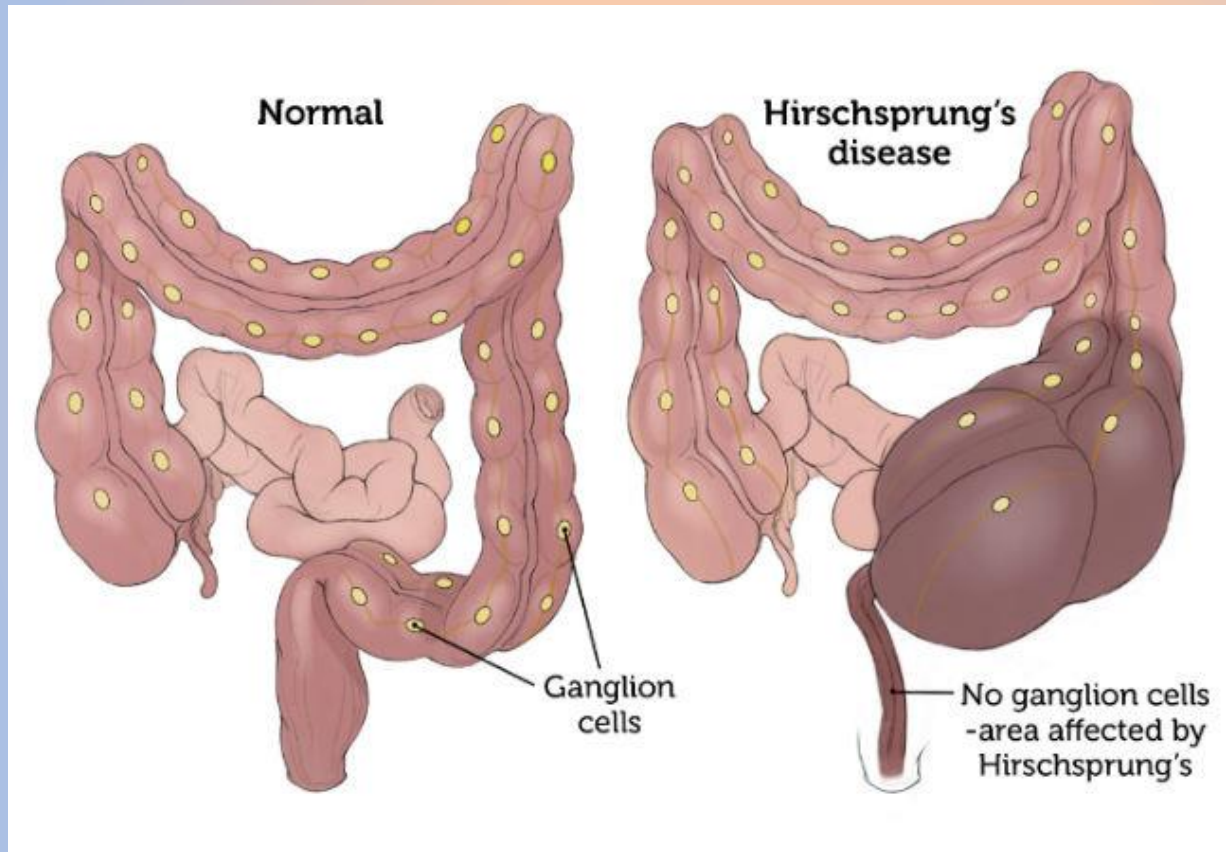
Duodenal atresia



Typical duodenum



Hirschsprung's Disease



<https://www.childrenshospital.org/conditions/hirschsprungs-disease>

Testing at Birth

Blood work:

- Karyotype to show chromosomes
- complete blood count with differential
- state metabolic screen will check thyroid function

Other tests

- all babies should get an **echocardiogram**
- hearing screen
- car seat challenge to make sure baby's oxygen stays normal while in car seat

Transient Myeloproliferative Disorder

Increased number of immature white blood cells

Most of the time this goes away

Puts the baby at risk for leukemia in the first 4 years of life

Needs evaluation by a hematologist

Birth to 1 Month

Monitoring of feeding and weight gain

Lactation consultation can be very helpful

<http://www.mobimotherhood.org/helping-babies-who-have-down-syndrome-learn-to-breastfeed.html>

<https://www.lli.org/breastfeeding-info/special-needs/>

<https://www.childrensmn.org/educationmaterials/childrensmn/article/15844/breastfeeding-an-infant-with-down-syndrome/>

Referral to early intervention

<https://ohioearlyintervention.org/>

Safe sleep



1 month to 1 year

Attention to feeding and weight gain

<https://www.cdc.gov/ncbddd/birthdefects/downsyndrome/growth-charts.html>

Repeat hearing test at 6 months old if newborn test was normal

Repeat thyroid testing at 6 months and 1 year

Evaluation by an eye doctor by age of 6 months

Blood count and test for iron deficiency at 1 year

Vaccines per usual schedule, some babies might qualify for RSV prevention



Concerns throughout Childhood

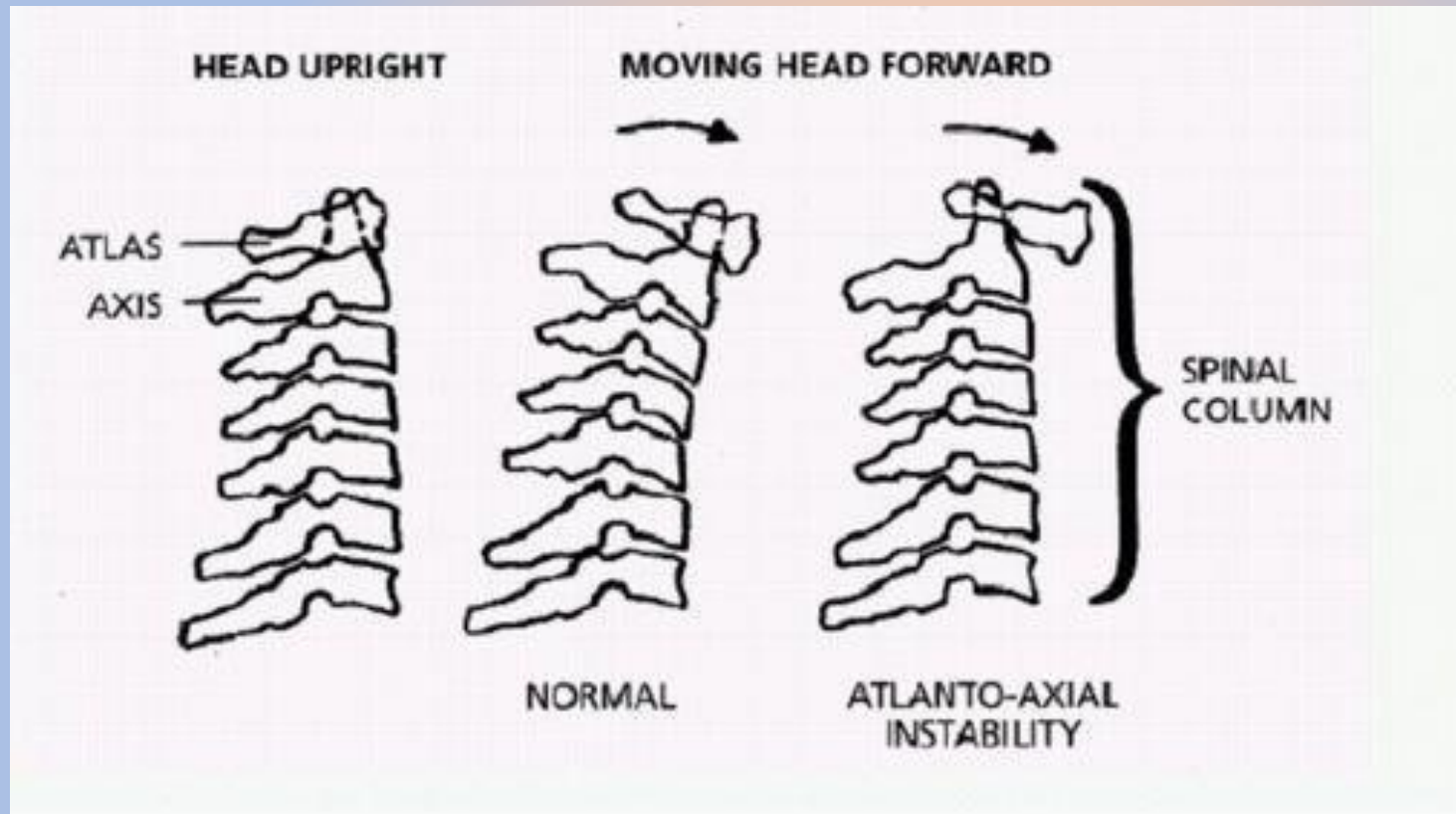
Obstructive Sleep apnea (OSA)

Overweight or obesity

Blood abnormalities

Atlantoaxial instability





<http://medcarepediatrics.blogspot.com/2013/04/atlanto-axial-instability-and-children.html>

Atlantoaxial Instability (AAI)

From the American Academy of Pediatrics

“Contact physician immediately for an x-ray of the neck in neutral position if your child has:

- Change in how he or she walks
- Change in how he or she uses arms/hands
- Change in bowel or bladder control
- Head stays tilted
- Neck Pain
- New onset weakness
- Decreased activity level or function

If the x-ray is abnormal or symptoms persist, the child should be referred as soon as possible to a pediatric neurosurgeon or pediatric orthopedic surgeon experienced in managing atlantoaxial instability.”

<https://www.healthychildren.org/English/health-issues/conditions/developmental-disabilities/Pages/Atlantoaxial-Instability-in-Children-with-Down-Syndrome.aspx>



AAI Precautions for All Children with Down syndrome

Avoid excessive flexion or extension of the neck during any, anesthetic, surgical or radiologic procedure to minimize risk of spinal cord injury

Avoid trampoline use unless it is part of a training program with appropriate supervision and safety measures

Participation in contact sports such as football, soccer, and gymnastics, places children at risk for spinal cord injury



Common Concerns

Autism

Attention Deficit Disorder

Behavior concerns

Regression

https://dsmig-usa.org/resources/Documents/21DSMIG_Regression%20in%20Persons%20with%20Down%20Syndrome_0928.pdf

Risk for abuse



Autoimmune diseases

Celiac disease

Thyroid disease

Type 1 diabetes

Alopecia

Juvenile Idiopathic Arthritis

Skin Conditions

Moya Moya

Dental care

Infectious diseases, especially pneumonia



1 Year to 5 Years

Monitor development, and screen for autism at 18 to 24 months

<https://www.kennedykrieger.org/stories/down-syndrome-and-autistic-spectrum-disorder-look-what-we-know>

Begin dental care

<https://www.ndss.org/resources/dental-issues-syndrome>

Sleep study at age 3-4 years, even if no sign of disordered breathing during sleep

Every check up monitor growth and assess for feeding issues

Yearly blood work to monitor blood count, iron deficiency and thyroid function

Yearly vision and hearing screening, review signs of atlantoaxial instability

Vaccines per usual schedule



Procedures can get more difficult at this age so consider social stories to prepare your child for a new experience

<https://sites.google.com/view/charlies-clinic/development/behavior#h.tngljx5m65ui>

This age is also a good time to talk to kids about body parts, appropriate touch, and privacy

These things will come up as potty training begins

<http://www.globaldownsyndrome.org/wp-content/uploads/2017/01/Potty-Time-Dr-Lina-Patel-Presentation.pdf>

At age 3yrs, responsibility for services and education transitions from early intervention to the school district and the child moves from an IFSP to an IEP



5 through 12 years old

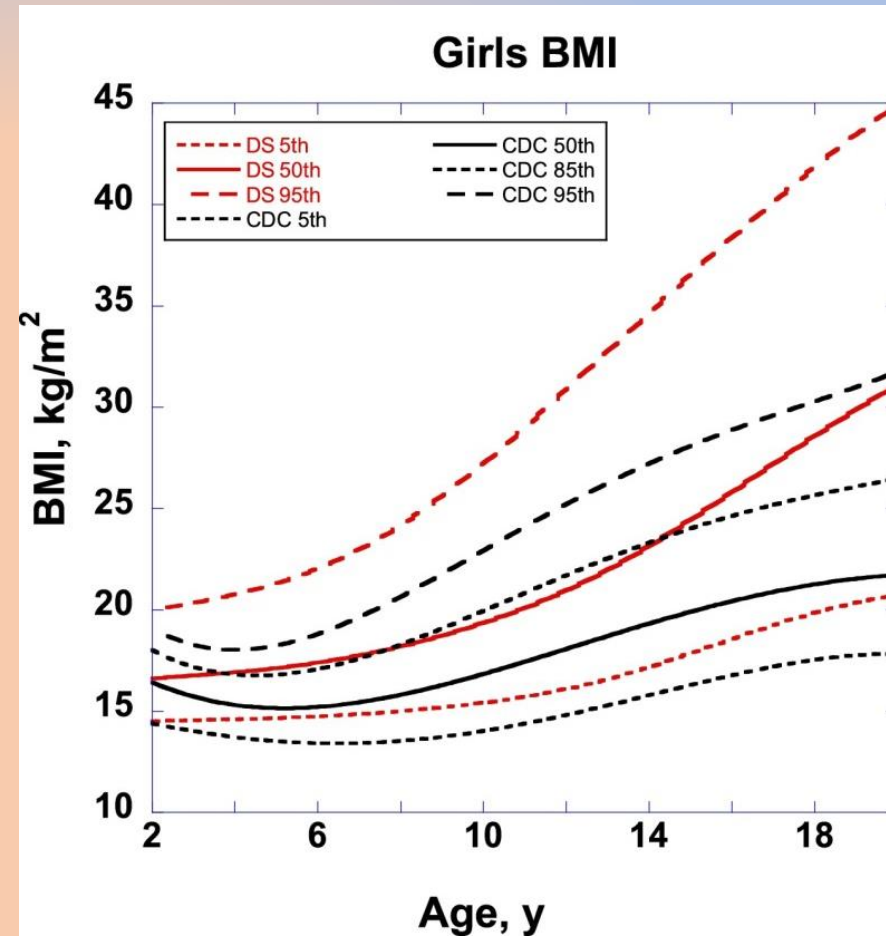
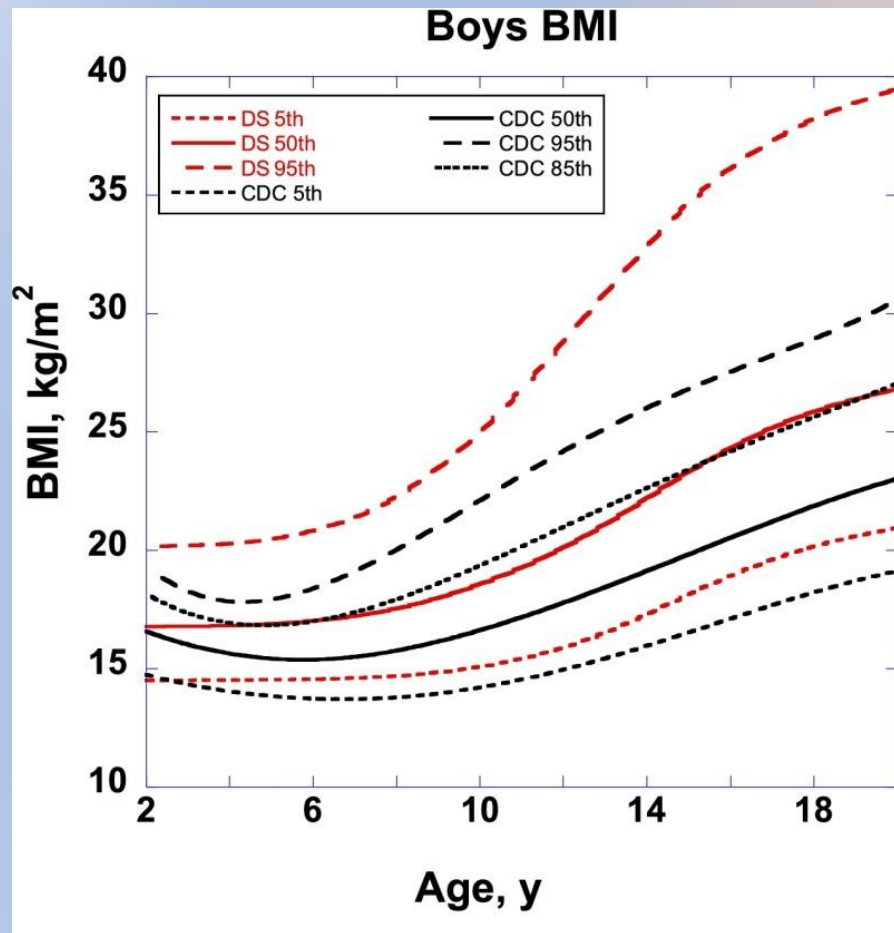
Monitor weight

Starting at 10 years old look at BMI on the chart not specifically for people with Ds to gauge weight status

Many teens and adults with Ds are overweight or obese and that puts them at risk for medical problems such as sleep apnea, type 2 diabetes, and joint problems

A healthy diet and regular exercise are important to prevent weight gain





[Pediatrics. 2016 Oct; 138\(4\): e20160541.](#)
doi: [10.1542/peds.2016-0541](https://doi.org/10.1542/peds.2016-0541)

Monitor development and behavior

Discuss transition to middle school

Encourage increasing ability in the bathroom

<https://adsresources.advocatehealth.com/bathing-and-showering-tips/>

<https://adsresources.advocatehealth.com/toilet-hygiene/>



Transition to middle school

Social media and internet use

Discuss puberty

Timing of puberty is not delayed

Period management

<https://adsresources.advocatehealth.com/menstrual-hygiene/>

<https://www.nytimes.com/wirecutter/reviews/thinx-period-panties/>



Story About How to Change My Pad



Terri Couwenhoven resources

Books:

**A Girls' Guide to Growing Up
Choices & Changes
in the Tween Years**

**The Boys' Guide to Growing Up
Choices & Changes
During Puberty**

Podcasts:

[Puberty and Sexuality Issues, Part 1](#)

[Puberty and Sexuality Issues, Part 2](#)



Assess hearing and vision

Yearly blood count, iron studies and thyroid functions

Vaccines per usual schedule



https://aap2.silverchair-cdn.com/aap2/content_public/journal/pediatrics/149/5/10.1542_peds.2022-057010/3/peds_2022057010supplementarydata.pdf

Thank You













TABLE 1

Medical Problems Common in Down Syndrome

Condition	%
Hearing problems	75
Vision problems	60–80
Nystagmus	3–33
Glaucoma	<1–7
Nasolacrimal duct occlusion	3–36
Cataracts	3
Strabismus	36
Refractive errors	36–80
Keratoconus	

Obstructive sleep apnea	50–79	
Otitis media with effusion		50–70
Congenital heart disease	40–50	
Feeding difficulty	31–80	
Respiratory infection	20–36	
Dermatologic problems	56	
Hypodontia and delayed dental eruption		23
Congenital hypothyroidism	2–7	
Antithyroid antibody positive (Hashimoto thyroiditis; incidence dependent on age)	13–39	
Hyperthyroidism	0.65–3	
Thyroid disease by adulthood		50
Gastrointestinal atresias	12	
Seizures	1–13	
Hematologic problems		
Anemia	1.2	
Iron deficiency	6.7	
Transient abnormal myelopoiesis		10
Leukemia	1	

Autoimmune conditions

Hashimoto thyroiditis	13–39	
Graves' disease	1	
Celiac disease	1–5	
Type 1 diabetes	1	
Juvenile idiopathic arthritis	<1	
Alopecia	5	
Symptomatic atlantoaxial instability	1–2	
Autism	7–19	
Hirschsprung disease	<1	
Moya Moya disease	Down syndrome 26 times greater in patients with Moyamoya	