Navigating Pediatric Healthcare for Children with Down Syndrome

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Disclosure Statement

- The information presented in this session is intended for educational purposes only.
- Content is based on the American Academy of Pediatrics (AAP) Health Supervision Guidelines for children and adolescents with Down Syndrome.
- This presentation is not a substitute for professional medical advice, diagnosis, or treatment.
- Families should consult their child's pediatrician and appropriate subspecialists for individualized recommendations.
- No conflicts of interest to disclose.

Overview

- AAP Health Supervision Guidelines
- Overview of Down Syndrome
- Age-Based Care: Infancy → Adolescence
- Practical Tools for Families
- Q&A

What is the American Academy of Pediatrics (AAP)?

The American Academy of Pediatrics is a professional organization for pediatricians.

It is the major organization for pediatric medicine in the United States, currently representing 67,000 physicians.

They support research through funding and publications, advocate for causes that are important for pediatricians, and establish best practices for the care of children and families.

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN™

AAP Health Supervision Guidelines for Children and Adolescents with Down Syndrome

The AAP publishes health supervision guidelines to help pediatricians care for children with Down syndrome.

First created in 1994, revised in 2001 and 2011, and most recently updated in 2022.

These guidelines provide preventative care recommendations: regular screenings, monitoring, and anticipatory guidance.

The goal is to identify medical problems early so children can receive timely treatment and support.

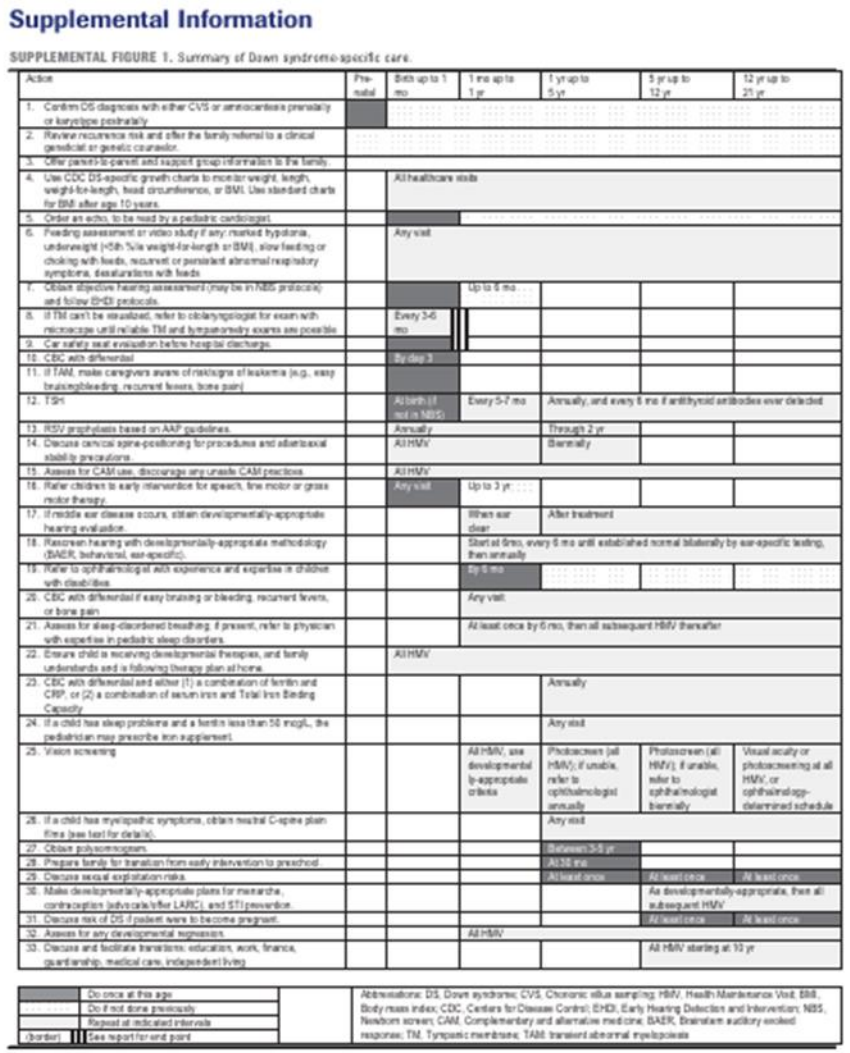
Takeaway: This is the roadmap pediatricians use to guide care for children with Down syndrome at every age.

https://publications.aap.org/pediatrics/article/149/5/e2022057010/186778/Health-Supervision-for-Children-and-Adolescents

AAP Health Supervision Guidelines for Children and Adolescents with Down Syndrome

Age Groups for Health Supervision

- Prenatal Period (before birth)
- Birth to 1 Month
- 1 Month to 1 Year
- 1 Year to 5 Years
- 5 Years to 12 Years
- 12 Years to 21 Years



Supplemental Information

SUPPLEMENTAL FIGURE 1. Summary of Down syndrome-specific care.

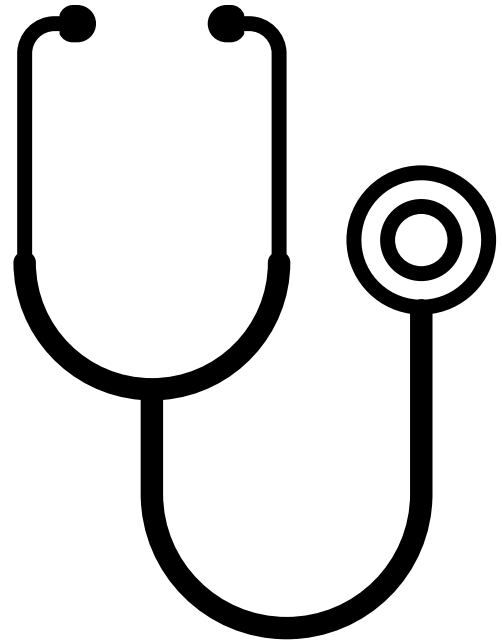
Action	Pre-natal	Birth up to 1 mo	1 mo up to 1 yr	1 yr up to 5 yr	5 yr up to 12 yr	12 yr up to 21 yr
1. Confirm DS diagnosis with either CVS or amniocentesis prenatally or karyotype postnatally.						
2. Review recurrence risk and offer the family referral to a clinical geneticist or genetic counselor.						
3. Offer parent-to-parent and support group information to the family.						
4. Use CDC DS-specific growth charts to monitor weight, length, weight-for-length, head circumference, or BMI. Use standard charts for BMI after age 10 years.		At healthcare visits				
5. Order an echo, to be read by a pediatric cardiologist.						
6. Feeding assessment or video study if any: marked hypotonia, underweight (<5th %ile weight-for-length or BMI), slow feeding or choking with feeds, recurrent or persistent abnormal respiratory symptoms, desaturations with feeds.		Any visit				
7. Obtain objective hearing assessment (may be in NBS protocols) and follow EHDI protocols.			Up to 6 mo			
8. If TM can't be visualized, refer to otolaryngologist for exam with microscope until reliable TM and tympanometry scans are possible.		Every 3-6 mo				
9. Car safety seat evaluation before hospital discharge.						
10. CBC with differential		By day 3				
11. If TAM, make caregivers aware of risk/signs of leukemia (e.g., easy bruising/bleeding, recurrent fevers, bone pain)						
12. TSH		At birth (if not in NBS)	Every 5-7 mo	Annually, and every 6 mo if antithyroid antibodies ever detected		
13. RSV prophylaxis based on AAP guidelines.		Annually		Through 2 yr		
14. Discuss cervical spine positioning for procedures and atlantoaxial stability precautions.		At HMV		Biennially		
15. Assess for CAM use, discourage any unsafe CAM practices.		At HMV				
16. Refer children to early intervention for speech, fine motor or gross motor therapy.		Any visit	Up to 3 yr			
17. If middle ear disease occurs, obtain developmentally-appropriate hearing evaluation.			When ear clear	After treatment		
18. Rescreen hearing with developmentally-appropriate methodology (BAER, behavioral, ear-specific).			Start at 6 mo, every 6 mo until established normal bilaterally by ear-specific testing, then annually			
19. Refer to ophthalmologist with experience and expertise in children with disabilities.			By 6 mo			
20. CBC with differential if easy bruising or bleeding, recurrent fevers, or bone pain			Any visit			
21. Assess for sleep-disordered breathing. If present, refer to physician with expertise in pediatric sleep disorders.			At least once by 6 mo, then all subsequent HMV thereafter			
22. Ensure child is receiving developmental therapies, and family understands and is following therapy plan at home.		At HMV				
23. CBC with differential and either (1) a combination of ferritin and CRP, or (2) a combination of serum iron and Total Iron Binding Capacity.				Annually		
24. If a child has sleep problems and a ferritin less than 50 mcg/L, the pediatrician may prescribe iron supplement.				Any visit		
25. Vision screening			At HMV, use developmentally-appropriate criteria	Photoscreen (all HMV); if unable, refer to ophthalmologist annually	Photoscreen (all HMV); if unable, refer to ophthalmologist biennially	Visual acuity or photoscreening at all HMV, or ophthalmologist-determined schedule
26. If a child has myelopathic symptoms, obtain neutral C-spine plain films (see text for details).				Any visit		
27. Obtain polysomnogram.				Between 3-5 yr		
28. Prepare family for transition from early intervention to preschool.				At 36 mo		
29. Discuss sexual exploitation risks.				At least once	At least once	At least once
30. Make developmentally-appropriate plans for menarche, contraception (advocate/offer LARC), and STI prevention.					As developmentally-appropriate, then all subsequent HMV	
31. Discuss risk of DS if patient were to become pregnant.					At least once	At least once
32. Assess for any developmental regression.			At HMV			
33. Discuss and facilitate transitions: education, work, finance, guardianship, medical care, independent living.					At HMV starting at 10 yr	

Do once at this age
Do if not done previously
Repeat at indicated intervals
(border) See report for end point

Abbreviations: DS, Down syndrome; CVS, Chorionic villus sampling; HMV, Health Maintenance Visit; BMI, Body mass index; CDC, Centers for Disease Control; EHDI, Early Hearing Detection and Intervention; NBS, Newborn screen; CAM, Complementary and alternative medicine; BAER, Brainstem auditory evoked response; TM, Tympanic membrane; TAM, transient abnormal myelopoiesis

Individualized Care and Health Supervision

- There is **no cure** for Down syndrome
- Every child needs an **individual treatment plan**
- Medical issues **change as a child grows**
- Health Supervision Guidelines are divided into **age groups** (newborn → early adulthood)
- Each age group highlights key issues by body system (heart, ears, thyroid, etc.)
- Some tests are **one-time only**, others must be **repeated regularly**



Common Medical Concerns in Down Syndrome

- **Intellectual disability**
- **Hearing loss** (~75%) & **ear infections** (50–70%)
- **Eye problems** (60–80%)
- **Obstructive sleep apnea** (50–79%)
- **Congenital heart disease** (~50%)
- **Thyroid disease** (24–50%)
- **Gastrointestinal problems**
- **Blood disorders**
- **Orthopedic concerns**
- **Autoimmune conditions**

A Positive Change in Care (AAP Guidelines, 2022)

- New recommendations emphasize respectful, supportive communication at diagnosis.
- Aligns with what family advocacy groups have been advocating for over many years.
- Celebrating your baby – congratulations first
- Respectful communication – use your baby's name, kind bedside manner
- Support – encourage parents, family, or loved ones to be present
- Accurate, up-to-date information – not opinions or outdated advice
- Person-first language – "child with Down syndrome"
- Connection – link to other parents, resources, and support groups
- Hope for the future – focus on life potential, not just challenges

TABLE 3 Communicating With Families[31]

At diagnosis, immediate advice remains pertinent regarding the need to:

- first, congratulate the family
- have infant present; refer to infant by name
- use a respectful bedside manner
- time discussion after labor is complete and as soon as diagnosis is suspected (not necessarily confirmed)
- have a support person present for mother, father, and family members as appropriate
- use a cohesive, physician-led team approach

Helpful discussion will include:

- up-to-date, accurate information
- a balanced approach rather than relying on personal opinions and experience
- person-first language (ie, child *with* Down syndrome)[32];
- connection to other parents and resource groups
- discussion of life potentials for people with Down syndrome

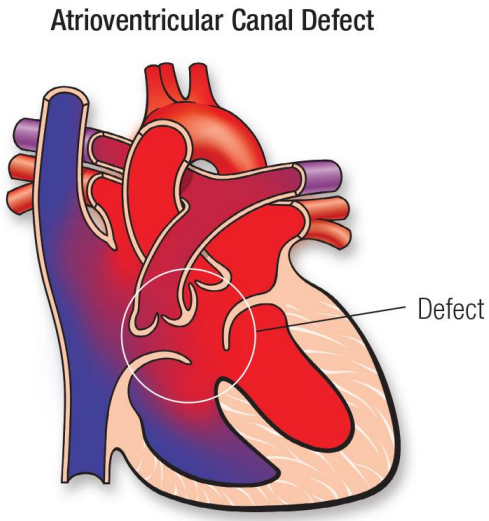
Share with families the interplay within families and individual perspectives:

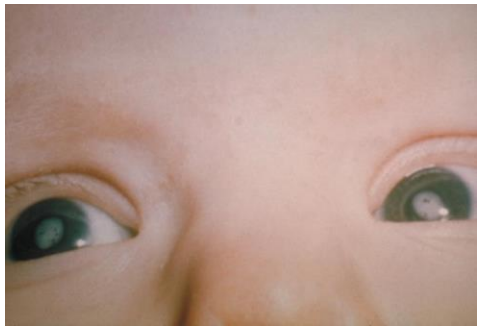
- individuals with Down syndrome: nearly 99% indicated that they were happy with their lives, and 97% liked who they are and encouraged health care professionals to value them, emphasizing that they share similar hopes and dreams as people without Down syndrome[33];
- parents: 79% felt their outlook on life was more positive because of people with Down syndrome[31];
- siblings: 88% felt that they were better people because of their siblings with Down syndrome[33];
- a majority of families report unanimous feelings of love and pride
- positive themes dominate modern families[34]

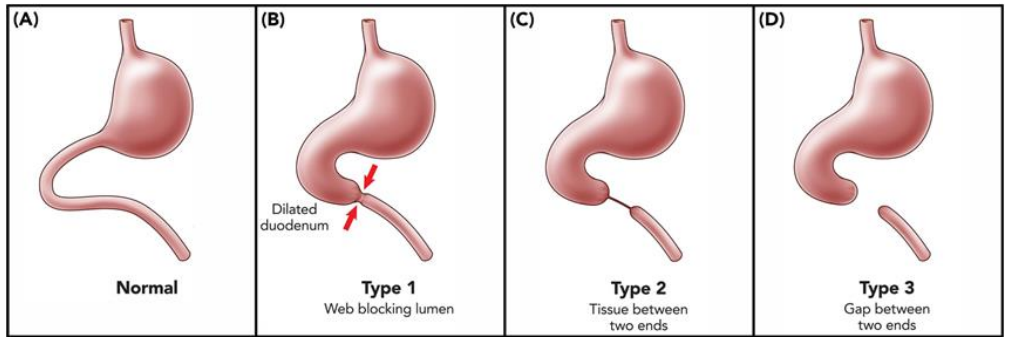
(1) Supervisions at Birth

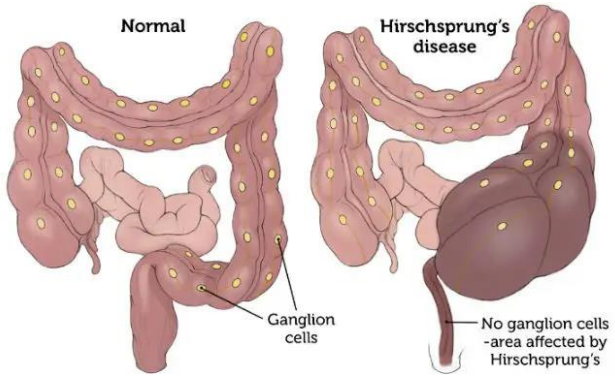
At birth, babies with Down syndrome will get a **thorough evaluation** to monitor for common medical issues, especially:

- **Heart problems**
- **Gastrointestinal problems** (duodenal atresia)
- **Feeding issues**
- **Eye problems** (such as cataracts)



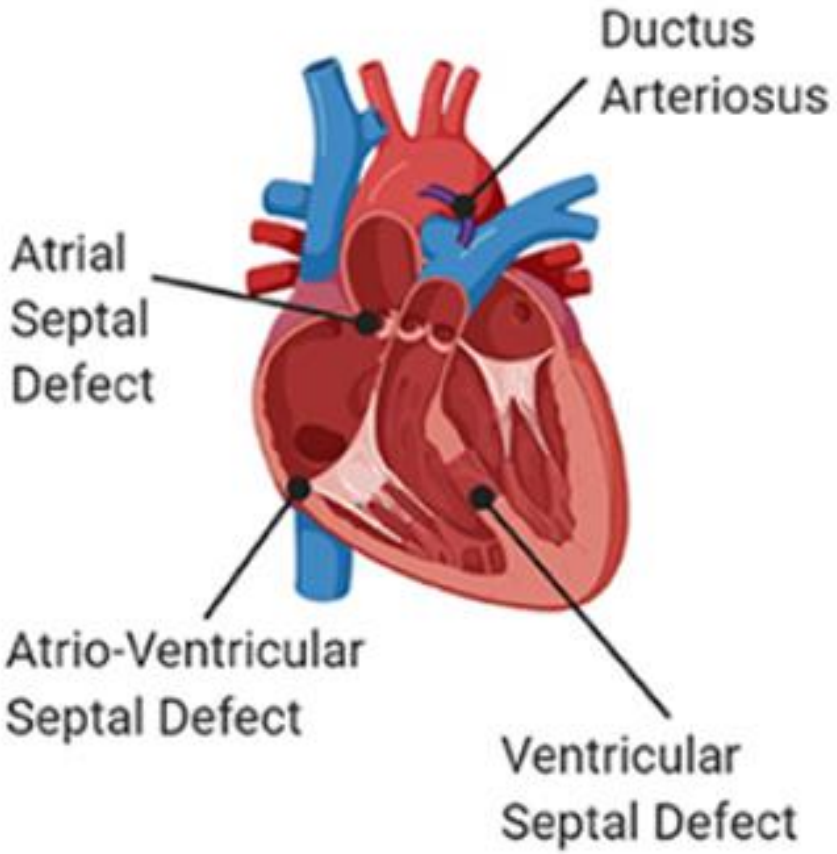






Cardiac Conditions

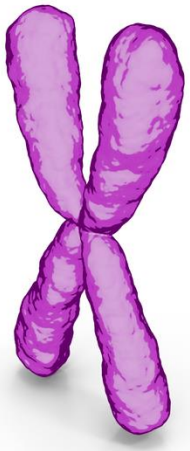
- Atrioventricular septal defect (AVSD) -> most common
- Ventricular septal defect (VSD)
- Atrial septal defects (ASD)
- Patent ductus arteriosus (PDA)

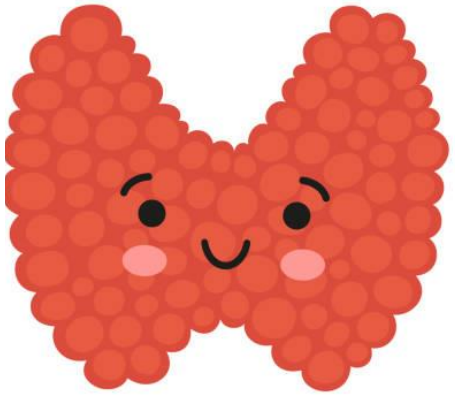


Testing

Blood Work

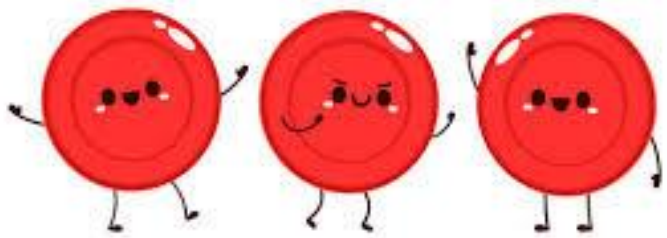
- Karyotype (chromosomes), if not already done
- Complete blood count (CBC) with differential
- Thyroid function test
  - 7% risk for hypothyroidism
  - 3% risk for hyperthyroidism





Other Tests

- Echocardiogram
- Hearing screen- risk for hearing loss
  - Fail: refer to ENT and early intervention
- Car seat challenge → check baby's oxygen levels while in car seat



Hematology in Down Syndrome

At Birth

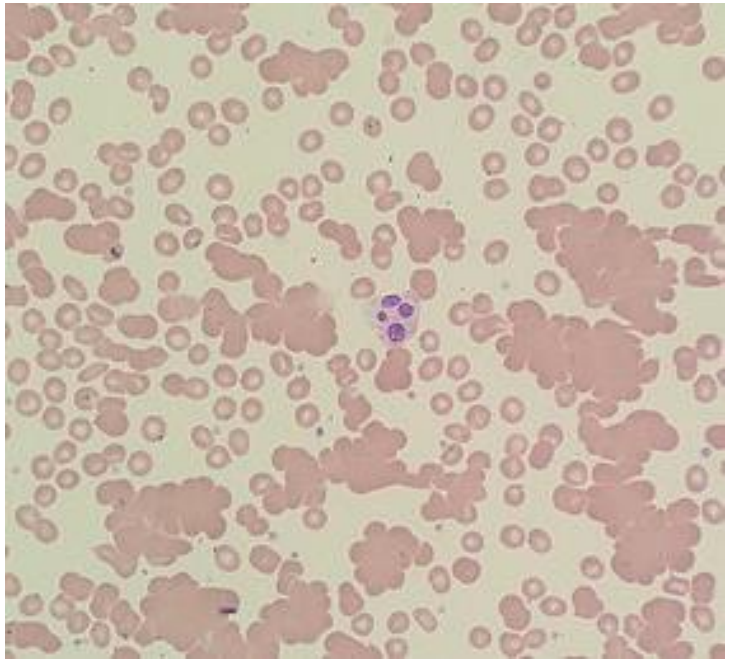
- Some babies are born with **extra immature blood cells**
- Usually goes away on its own
- Sometimes needs follow-up with a blood doctor (hematologist)

As Children Grow

- Kids with Down syndrome are more likely to have **low iron (anemia)**
- Blood tests at **1 year old and every year after** help check for anemia
- Low iron can cause **tiredness, poor sleep, or behavior changes**

Leukemia Risk

- Slightly higher risk (**about 1 in 100**) compared to other kids
- Important to watch for:
  - Easy bruising or bleeding
  - Bone or joint pain
  - Extreme tiredness



(2) Supervision from Birth to 1 Month

Feeding & Growth

- Monitor weight gain
- Babies with Down syndrome have their **own growth curves**

Early Intervention

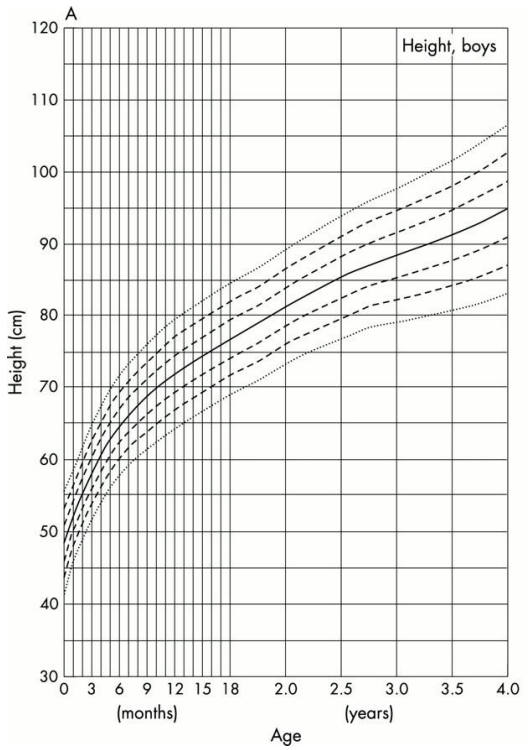
- Referral right away for developmental supports

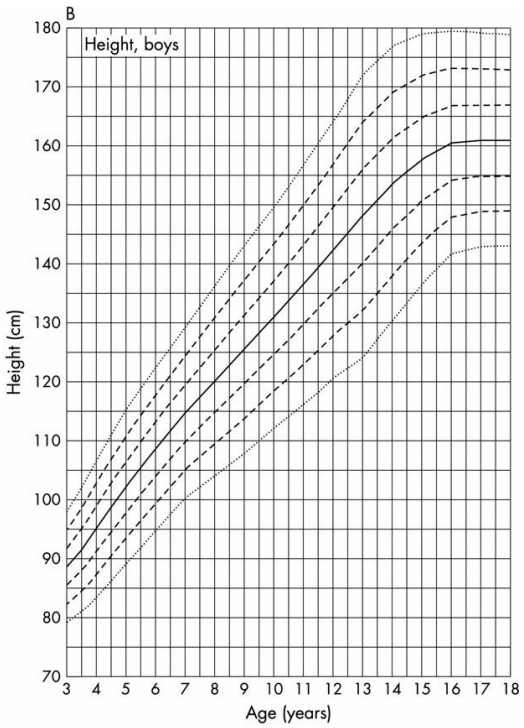
Respiratory Health

- Discuss increased susceptibility to infections

Cervical Spine Safety

- Guidance on safe handling and positioning





Why More Infections in Down Syndrome?

Respiratory Infections

Smaller airway and large tongue → easier to get blocked

Low muscle tone → harder to clear mucus/cough effectively

Immune system differences → more colds and pneumonia

Heart conditions (if present) → can make breathing problems worse

Sleep apnea → lowers oxygen, adds stress on lungs

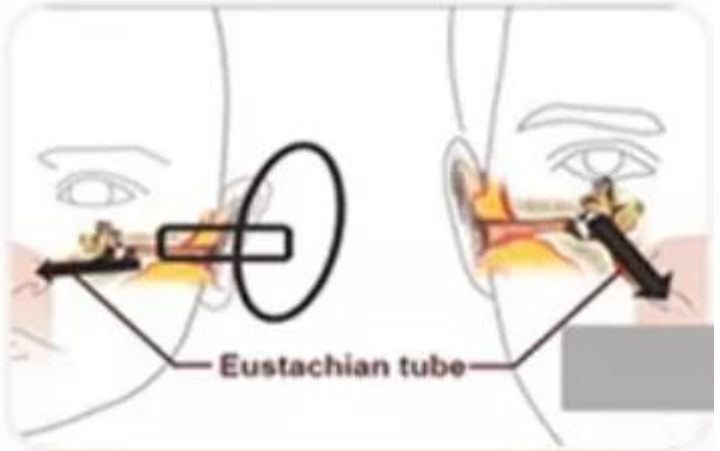
Ear Infections

Narrow ear canals → fluid builds up more easily

Eustachian tube problems → poor drainage behind the ear

Weaker immune response → more frequent and longer-lasting infections

Repeated infections → can cause hearing loss if untreated

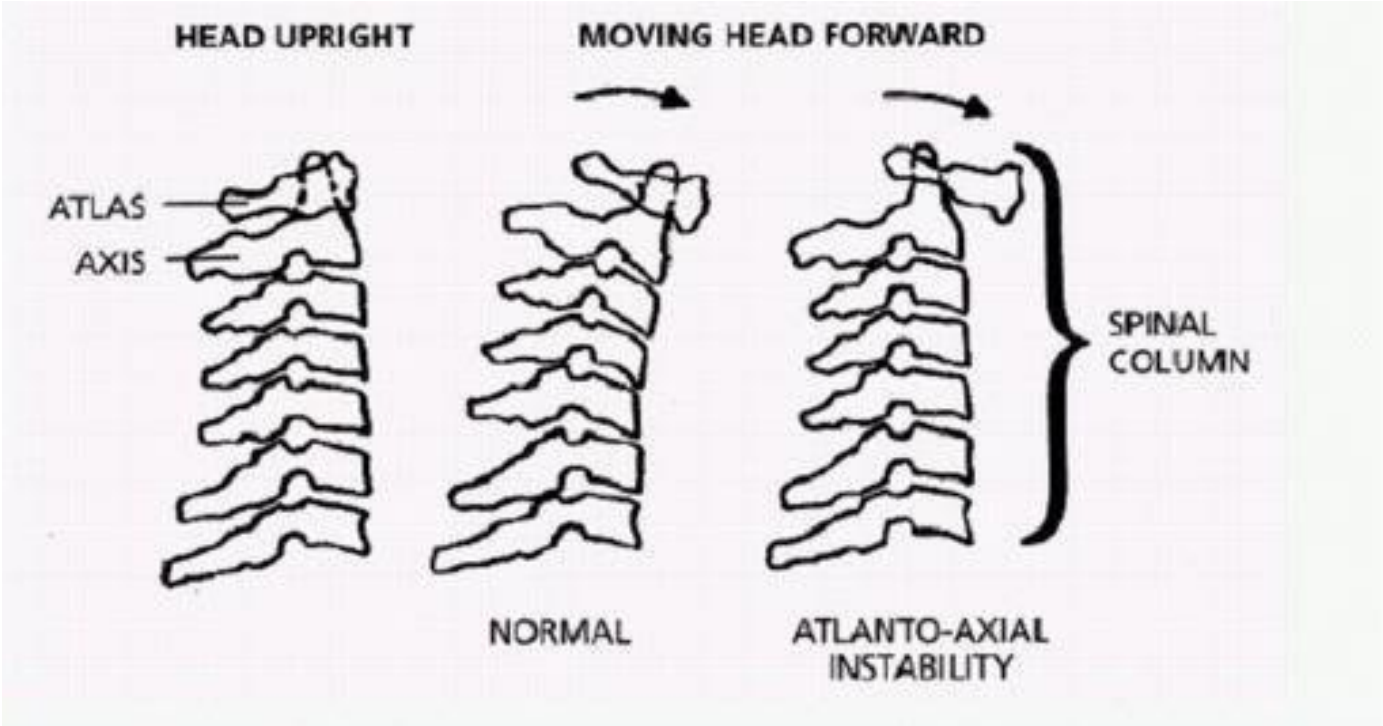


Anatomy Differences

- Small low set ears
- Narrow ear canals
- Narrow and horizontal eustachian tube
- Large adenoids
- Smaller nasal cavity

Cervical Spine Safety & Atlantoaxial Instability (AAI)

- Extra flexibility in the neck bones can cause instability
- Risk of spinal cord injury with excessive neck extension or flexion
- Precautions needed during anesthesia, surgery, or imaging



AAI Precautions for All Children with Down Syndrome

Neck Safety
- Avoid excessive flexion or extension during anesthesia, surgery, or imaging
- Prevents risk of spinal cord injury

Trampolines: Avoid use unless part of a supervised training program with safety measures

Sports
- Contact sports (football, soccer, gymnastics) increase the risk of spinal cord injury
- Discuss safe activity options with your child's doctor

Signs: Neck pain, weakness, abnormal reflexes, change in bowel/bladder function, head tilt
  - Contact your child's doctor
  - Imaging should be obtained if there are symptoms

(3) Supervision at 1 Month to 1 Year

Feeding & Growth
- Monitor feeding and weight gain

Hearing & Vision
- Repeat hearing test at 6 months (if newborn screen was normal)
- Eye doctor evaluation by age 6 months

Thyroid
- Test at 6 months and 1 year

Blood Work
- CBC and iron studies at 1 year

Sleep & Neurology
- Discuss signs of sleep apnea
- Monitor for seizures, infantile spasms, or AAI
  - Up to 13% may have seizures
    - Many in 1st year of life

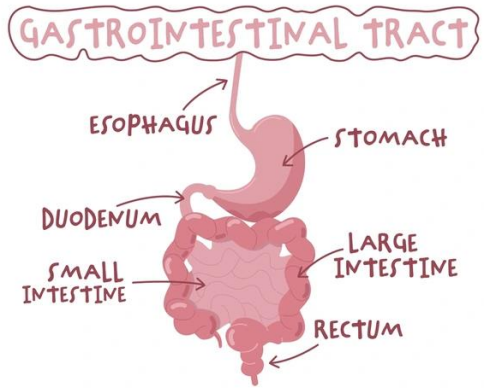
Vaccines
- Follow regular schedule
- Some babies may qualify for **RSV prevention**

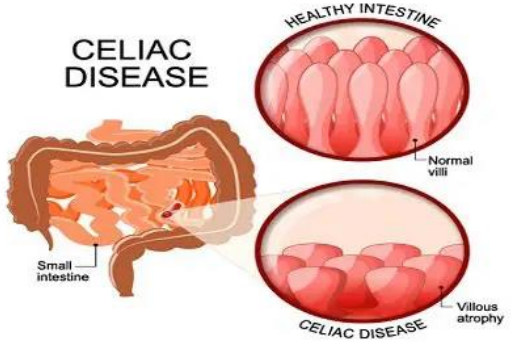
Feeding Concerns and Gastrointestinal Concerns

- GERD(Gastroesophageal Reflux Disease)
- Swallowing difficulties (dysphagia)
- Low muscle tone (hypotonia)
- Large tongue (macroglossia)

- Referral to feeding specialist if:
  - Marked hypotonia
  - Underweight
  - Slow feeding
  - Choking with feeds
  - Persistent respiratory symptoms
  - Low oxygen levels with feeds

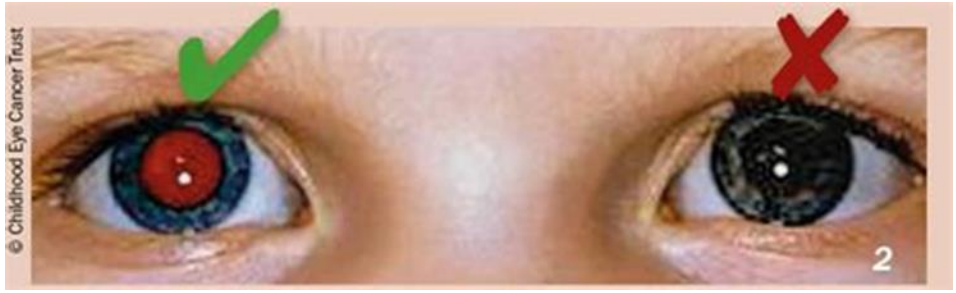
- Celiac Disease
  - Blood test (Celiac IgA) at age 2–3 years
  - Test earlier if symptoms present:
    - Diarrhea
    - Constipation
    - Slow growth
    - Anemia
    - Abdominal pain
- Constipation

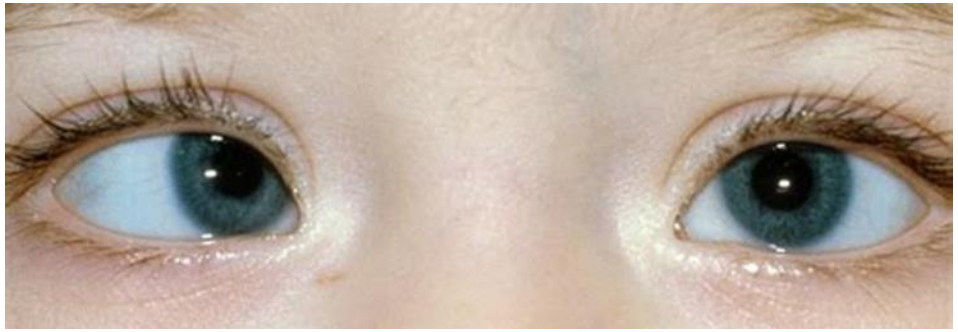




Ocular Conditions

- See Ophthalmologist by 6 months
- Screen yearly at pediatrician's office
- Evaluate every 2 years by ophthalmologist
- 50% risk of refractive errors
- Start screening early
- Strabismus (misaligned eyes), Glaucoma (increased pressure)





(4) Supervision at 1 Year to 5 Years

- **Development**
  - Monitor milestones
  - Autism screening at **18–24 months**
- **Dental Care**
  - Begin dental visits
- **Sleep**
  - Sleep study at age **3–4 years**, even without symptoms
- **Growth & Nutrition**
  - Monitor growth, feeding issues, and stooling patterns
- **Blood Work**
  - Yearly CBC, iron studies, and thyroid function
- **Vision & Hearing**
  - Annual screening
- **Vaccines**
  - Per standard schedule
- **Neck Safety**
  - Review signs of atlantoaxial instability (AAI)

Development and Milestones

Developmental Strengths

- Visual learning and memory
- Early social development and motivation
- Word reading
- Strong desire to connect with others

Common Challenges

- Speech and language delays (often more affected than motor skills)
- Intellectual disability: usually **mild (IQ 50–70) to moderate (IQ 35–50)**
- Continued development at a slower rate, but progress continues into adulthood

Support & Services

- **Early Intervention (0–3 years)** – *Early Start in California* under "established risk"
- **After age 3** – School-based services (**IEP**) and Regional Center supports
- Multidisciplinary therapies (speech, occupational, physical, feeding)

Autism & Down Syndrome

Prevalence
- Present in **16–18%** of individuals with Down syndrome

Core Features of Autism
- Social communication differences
- Repetitive and restricted interests/behaviors

DS + ASD Presentation
- May look **different** than autism without genetic causes
- Often greater challenges with **language and cognitive skills** compared to DS alone

Support
- Behavioral therapies (**Applied Behavior Analysis**) have been shown to be effective

Obstructive Sleep Apnea (OSA) in Down Syndrome

Why it happens:

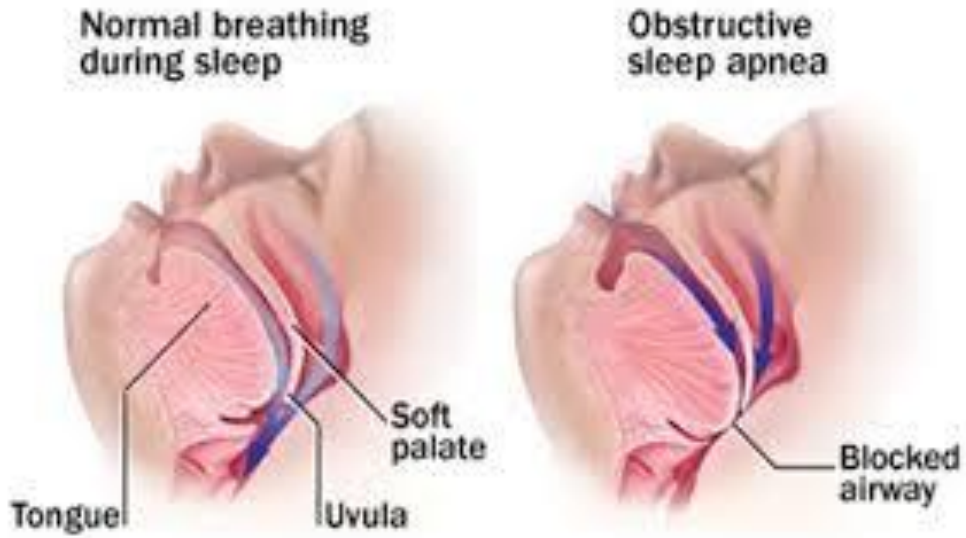
- Smaller upper airway
- Low muscle tone
- Enlarged tonsils/adenoids

Common Symptoms:

- Snoring
- Heavy or noisy breathing
- Unusual sleep positions
- Night awakenings
- Daytime sleepiness or behavior changes

Screening & Monitoring:

- **Sleep study recommended at age 3–5 years**, even if no symptoms
- Repeat studies if symptoms develop later



(5) Supervision at 5 to 12 Years

Development & Learning

- Monitor school progress, attention, and behavior
- Screen for **ADHD, anxiety, and mood issues**
- Support with IEP/504, focus on **literacy and social skills**

Dental Care- Regular visits every 6 months

Sleep

- Monitor for snoring, restless sleep, or daytime fatigue
- Repeat sleep study if symptoms develop

Growth & Nutrition

- Track weight and BMI (risk of **obesity & metabolic syndrome**)
- Monitor for **constipation and celiac symptoms**

Blood Work- Annual CBC, iron studies, and thyroid function

Vision & Hearing- Annual screening, ENT follow-up for chronic ear issues

- **Orthopedics-** Low muscle tone and lax joints
- Monitor for **hip instability, patellar dislocation, flat feet, scoliosis**

Vaccines- Continue per standard schedule

Neck Safety- Review signs of **atlantoaxial instability (AAI)**, especially before sports

ADHD in Children with Down Syndrome

How common?

- Up to **40% of children with Down syndrome** may also have ADHD

Why it's hard to diagnose

- ADHD symptoms can overlap with:
  - Sleep problems
  - Vision or hearing issues
  - Thyroid disease
  - Communication difficulties
  - Autism

Treatment Options

- **Behavioral strategies first**: parent training, structured routines, therapy (e.g., PCIT, Triple P)
- **Medication**: sometimes used if symptoms are severe, but less common in Down syndrome because of side effects and limited research
- Ongoing studies are exploring **safe and effective use** of stimulant medications

Key takeaway for parents:

- If your child struggles with focus, impulsivity, or hyperactivity, talk with your doctor
- Rule out other medical causes first (sleep, thyroid, hearing, vision)
- Treatment is individualized -> no "one-size-fits-all"

(6) Supervision at 12 to 21 Years

Adolescent Development:

- Puberty: timing, self-care, hygiene, and **sexual health education**
- Screen for **depression, anxiety, and behavior changes**
- Promote independence in daily living skills

Transition Planning

- Begin by **age 12**: plan for transition to adult medical care, education, employment, and community living
- Coordinate with school (IEP → transition plan at 16)
- Encourage self-advocacy and decision-making

Dental Care- Ongoing dental visits every 6 months

Sleep

- Monitor for **obstructive sleep apnea**
- Repeat sleep study if new symptoms

Growth & Nutrition

- Watch for **obesity, metabolic syndrome, and thyroid disease**
- Continue celiac screening if symptoms

Blood Work- Annual CBC, iron studies, thyroid function

Vision & Hearing- Annual screening

Orthopedics- Continue monitoring for **hip, scoliosis, or joint issues**

Vaccines- Ensure full **adolescent series**

Neck Safety & Sports- Ongoing precautions for **AAI** with sports and anesthesia

Mental Health in Children & Teens with Down Syndrome

Common Concerns

- **Anxiety-** Can look like obsessive routines, worries, or specific fears
- **Depression-** May show as loss of interest, irritability, or withdrawal
- **Behavioral challenges-** Oppositional or difficult behaviors may increase with stress
- **Regression (rare)**
  - "Down Syndrome Disintegrative Disorder"
  - Usually, in adolescence, more common in girls
  - Sudden decline in language, communication, and social skills

What Parents Can Do

- Watch for **changes in sleep, appetite, mood, or school performance**
- Seek support early: therapy, counseling, medical evaluation -> rule out medical cause
- Create routines, positive social opportunities, and stress management strategies

Key takeaway: Children and teens with Down syndrome can experience **mental health challenges just like other kids** -> early recognition and support make a big difference.

Summary of Recurring Screenings

- **Vision & Hearing** – every year
- **Growth monitoring** – use DS-specific growth charts
- **Dental exam** – every 6 months
- **Thyroid testing** – every year
- **CBC with differential + iron studies (Ferritin + CRP OR Iron + TIBC) – annually**
- **One-time / age-specific tests:**
  - **Celiac IgA** – at 2–3 years (earlier if symptoms)
  - **Sleep study** – by age 3–5 years (earlier if symptoms of OSA)

Beyond Medical Care

- **Social and school success** are also important
- Often have strong **social skills** with positive peer models
- Each person has **unique strengths and talents**
  - Acknowledging strengths builds confidence.
- People with Down syndrome are **more like their peers than different**

Social Stories for Children & Teens with Down Syndrome

What are Social Stories? Short, simple, **visual stories** that explain what to expect in a situation

- Help reduce **anxiety** and improve **cooperation**

Why They Help

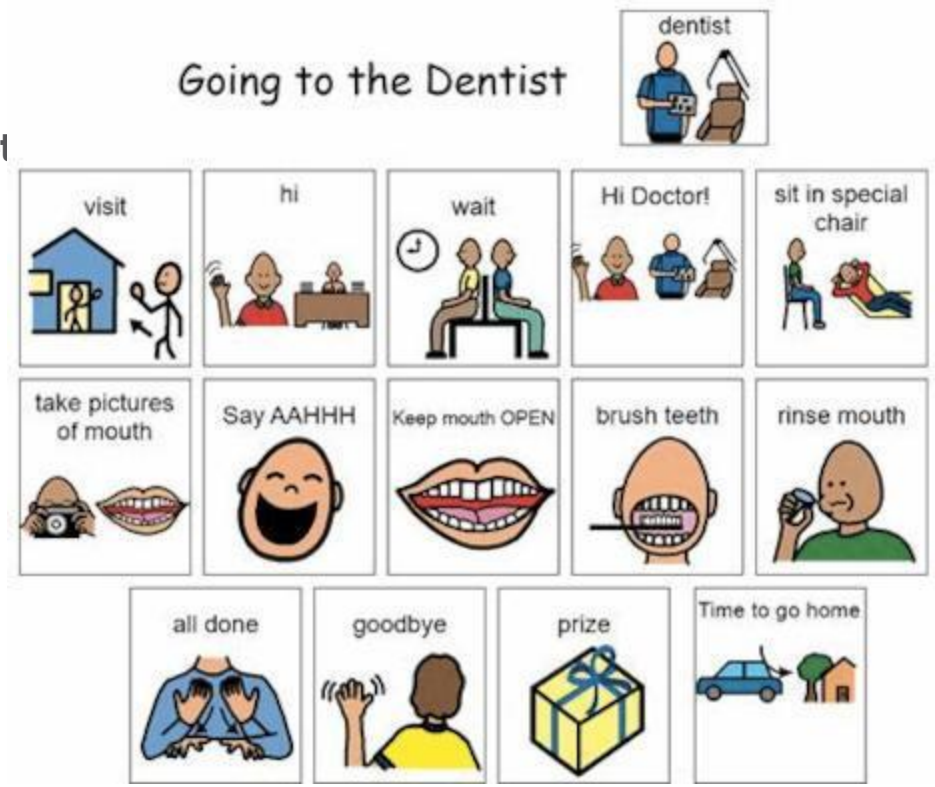
- Children with Down syndrome often learn best with **pictures and repet**
- Make unfamiliar routines more **predictable and less stressful**

When to Use (Across All Ages)

- **Medical care**: doctor visits, blood draws, dental work, surgeries
- **School transitions**: starting a new grade, meeting a new teacher
- **Daily living**: potty training, bedtime routines, chores, self-care
- **Social situations**: birthday parties, making friends

Tips for Parents

- Use **real photos or symbols** when possible
- Keep language **positive, short, and clear**
- Practice before the event and **repeat often**
- Pair stories with **role-play or modeling**



Medical Passports & Summaries

What is it?

- A **portable medical summary** that keeps all your child's important health information in one place
- Easy to share with **doctors, schools, therapists, and emergency providers**

Why it's important in Down Syndrome

- Children often see **multiple specialists** (heart, ENT, endocrinology, therapies)
- Prevents missed information and reduces stress during emergencies or hospital visits
- Helps new providers quickly learn about your child's **strengths, challenges, and supports**

What to include

- Diagnoses, surgeries, allergies, and medications
- Contact info for all providers
- Medical equipment and insurance details
- Family notes: strengths, communication needs, things to avoid, ways to help

Example:

Portable Medical Summary

Children's Hospital at Dartmouth-Hitchcock

PAGE 1 Date completed _____

CHILD'S NAME: _____ DATE OF BIRTH: _____ AGE: _____

CHILD'S NICKNAME: _____ HEIGHT: _____ WEIGHT: _____

ADDRESS AND CONTACT INFORMATION

Parents/Guardians: _____ Relationship: _____

Address: _____ City: _____ State/zip: _____

Home phone: _____ Cell phone: _____

Primary language/Communication: _____

Important things to know about my child: _____

MEDICAL INFORMATION

Diagnosis (es): _____ Age when diagnosed: _____

_____ Age when diagnosed: _____

_____ Age when diagnosed: _____

ALLERGIES/SENSITIVITIES	Reactions	ALLERGIES/SENSITIVITIES	Reactions

MEDICATIONS	Dose	Time	OTC Medications/Supplements

Portable Medical Summary

Children's Hospital at Dartmouth-Hitchcock

PAGE 2 Date completed _____

HEALTHCARE AND OTHER PROVIDERS

Primary Care Provider: _____ Phone: _____
Fax: _____

Specialty Provider: _____ Phone: _____
Fax: _____

Specialty Provider: _____ Phone: _____
Fax: _____

Other Provider: _____ Phone: _____
Fax: _____

Other Provider: _____ Phone: _____

HOSPITALIZATIONS

Date	Surgeries/Procedures/Illnesses	Hospital/Doctor

MEDICAL EQUIPMENT/SUPPLIES	Provider	Contact Info.

HEALTH INSURANCE - *PRIMARY*

Name: _____
Group #: _____
ID #: _____
Phone: _____
Fax: _____

HEALTH INSURANCE - *SECONDARY*

Name: _____
Group #: _____
ID #: _____
Phone: _____
Fax: _____

Portable Medical Summary

Children's Hospital at Dartmouth-Hitchcock

PAGE 3 Date completed _____

GETTING TO KNOW MY CHILD

NAME: _____ NICKNAME: _____

DATE OF BIRTH: _____ TODAY'S DATE: _____

A little about my child

My child's strengths (things that are easy for my child)

My child's challenges (examples: communication, feeding, learning, mobility, socialization, energy, behavior)

My child's life in the community (examples: school, childcare, place of worship, favorite places)

Ways you can help my child (examples: speak quietly, give choices, do not talk about scary things)

Things to avoid (examples: food, activities, and procedures)

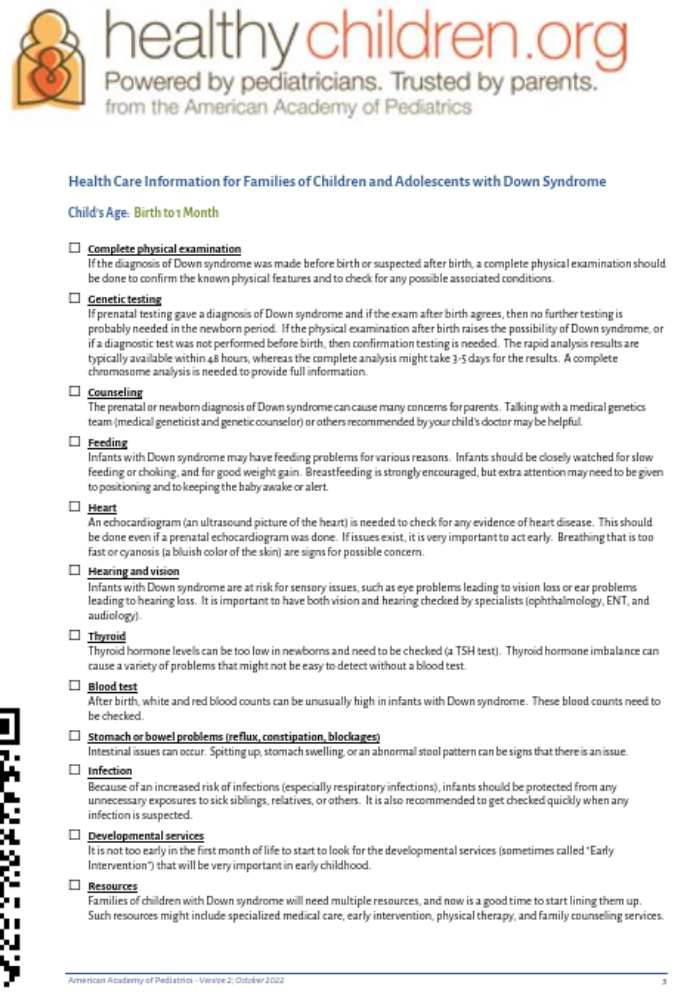
Other important information

Tip: Keep laminated copies of the Portable Medical Summary on hand, or take a picture and keep a copy on mobile devices.

UCDAVIS HEALTH | MIND INSTITUTE

Family-Friendly Guidelines: HealthyChildren.org

- Provides the AAP Down syndrome guidelines in a checklist format for families.
- Easy to understand and organized by child's age.
- Covers recommended screenings, medical checks, and developmental supports.
- Helps parents track what to expect at each stage and what to ask at appointments.
- https://www.healthychildren.org/English/health-issues/conditions/developmental-disabilities/Pages/Children-with-Down-Syndrome-Health-Care-Information-for-Families.aspx


healthychildren.org
Powered by pediatricians. Trusted by parents.
from the American Academy of Pediatrics

Health Care Information for Families of Children and Adolescents with Down Syndrome

Child's Age: Birth to 1 Month

☐ **Complete physical examination**
If the diagnosis of Down syndrome was made before birth or suspected after birth, a complete physical examination should be done to confirm the known physical features and to check for any possible associated conditions.

☐ **Genetic testing**
If prenatal testing gave a diagnosis of Down syndrome and if the exam after birth agrees, then no further testing is probably needed in the newborn period. If the physical examination after birth raises the possibility of Down syndrome, or if a diagnostic test was not performed before birth, then confirmation testing is needed. The rapid analysis results are typically available within 48 hours, whereas the complete analysis might take 3-5 days for the results. A complete chromosome analysis is needed to provide full information.

☐ **Counseling**
The prenatal or newborn diagnosis of Down syndrome can cause many concerns for parents. Talking with a medical genetics team (medical geneticist and genetic counselor) or others recommended by your child's doctor may be helpful.

☐ **Feeding**
Infants with Down syndrome may have feeding problems for various reasons. Infants should be closely watched for slow feeding or choking, and for good weight gain. Breastfeeding is strongly encouraged, but extra attention may need to be given to positioning and to keeping the baby awake or alert.

☐ **Heart**
An echocardiogram (an ultrasound picture of the heart) is needed to check for any evidence of heart disease. This should be done even if a prenatal echocardiogram was done. If issues exist, it is very important to act early. Breathing that is too fast or cyanosis (a bluish color of the skin) are signs for possible concern.

☐ **Hearing and vision**
Infants with Down syndrome are at risk for sensory issues, such as eye problems leading to vision loss or ear problems leading to hearing loss. It is important to have both vision and hearing checked by specialists (ophthalmology, ENT, and audiology).

☐ **Thyroid**
Thyroid hormone levels can be too low in newborns and need to be checked (a TSH test). Thyroid hormone imbalance can cause a variety of problems that might not be easy to detect without a blood test.

☐ **Blood test**
After birth, white and red blood counts can be unusually high in infants with Down syndrome. These blood counts need to be checked.

☐ **Stomach or bowel problems (reflux, constipation, blockages)**
Intestinal issues can occur. Spitting up, stomach swelling, or an abnormal stool pattern can be signs that there is an issue.

☐ **Infection**
Because of an increased risk of infections (especially respiratory infections), infants should be protected from any unnecessary exposures to sick siblings, relatives, or others. It is also recommended to get checked quickly when any infection is suspected.

☐ **Developmental services**
It is not too early in the first month of life to start to look for the developmental services (sometimes called "Early Intervention") that will be very important in early childhood.

☐ **Resources**
Families of children with Down syndrome will need multiple resources, and now is a good time to start lining them up. Such resources might include specialized medical care, early intervention, physical therapy, and family counseling services.

Questions?

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