

## Down Syndrome

### *For Healthcare Providers*

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools ([www.negenetics.org](http://www.negenetics.org)).

### Physical characteristics and/or symptoms

*Note: not all people with Down syndrome will have all of these features.*

- Hypotonia
- Characteristic facial features:
  - Nose
    - Flat bridge of the nose
  - Eyes
    - Upslanting palpebral fissures
    - Epicanthal folds
    - Brushfield spots
  - Mouth
    - Tends to be small and the roof of mouth may be shallow
    - Tongue often protrudes and appears large in relation to the mouth due to low muscle tone.
  - Teeth
    - May come in late, be small, and appear in an unusual order
  - Ears
    - Small, helices fold over, absent ear lobes, and set lower on head
    - Smaller ear passages that can lead to more frequent ear infections
  - Head shape
    - Smaller than normal
  - Neck
    - Appears shorter with excess folds of skin on the back of the neck
- Stature:
  - Average adult female height is 4 feet 9.
  - Average adult male height is 5 feet 2 inches.
- Hands and Feet:
  - Small with short fingers

- Single crease extending across the palm of one or both hands
- 5<sup>th</sup> finger clinodactyly
- Toes have a gap between 1<sup>st</sup> and 2<sup>nd</sup> toes
- Chest abnormality
- Skin
  - Mottling is common
- Hair
  - Thin, soft, and/or sparse

## Recommended Routine Surveillance

- Management of hypotonia
- Cardiac evaluation
- Measure TSH annually
- Obtain hemoglobin annually
- Discuss symptoms related to celiac disease.
- Aggressive treatment of respiratory tract infections
- Vision and hearing evaluation
- Evaluation of respiratory issues and sleep apnea
- Cervical spine positioning precautions

## Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Emergencies should be handled as with any child.
- If seizures are present, the following seizure action plan may be useful:  
<https://www.epilepsy.com/learn/managing-your-epilepsy/seizure-action-plans>

## Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Cardiologist
  - Cardiac abnormalities
- Dentist
  - Dental anomalies

- Developmental evaluation:
  - Acquired mitral and aortic valvular disease
  - Speech therapy
  - Physical therapy
  - Occupational therapy
- ENT
  - Hearing loss
  - Otitis media
- Endocrinologist
  - Hypothyroidism
- Gastroenterologist
  - Feeding problems
  - Constipation
  - Reflux
  - Celiac disease
- Geneticist / Genetic Counselor:
  - Diagnosis
  - Coordination of care
  - Genetic risk for family
  - Clinical trials
- Hematologist
  - Transient myeloproliferative disorder (TMD)
  - Leukemia
- Neurology
  - Monitor seizures
- Nutritionist
  - Management of diet and exercise program
- Ophthalmologist
  - Cataracts
  - Strabismus
  - Nystagmus
- Orthopedists
  - Atlantoaxial instability
- Pulmonologist
  - Airway anomalies
  - Breathing issues
- Sleep specialist
  - Sleep apnea
- Behavior and psychiatric evaluation

## Sample Forms

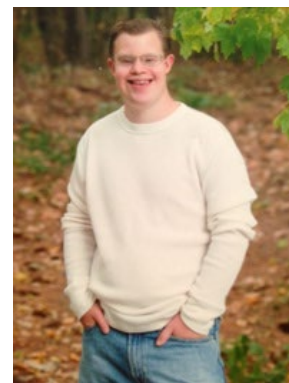
- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

*My patient \_\_\_\_\_ has been diagnosed with Down syndrome. This condition is characterized by intellectual disability, characteristic facial features, hypotonia, and cardiac abnormalities. Medical complications with Down syndrome include management of cardiac abnormalities, gastrointestinal difficulties, and hearing and vision problems. Because of these, \_\_\_\_\_ needs the following accommodations.*

## Seven Important Aspects of School Life

“[Down Syndrome at a Glance](#)” will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions



## Resources

### American Academy of Pediatrics (AAP) - Health Supervision Guidelines

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

### Medline Plus

<https://medlineplus.gov/genetics/condition/down-syndrome/>

Additional resources including support group information can be found on the main website.