



CHCBS brief + Med info

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Brief [REDACTED] Hearing

[REDACTED] is a seven-year-old child with Non Mosaic Downs Syndrome¹ and a host of co-morbid medical conditions for which he needs daily skilled treatment. These conditions are not stable and even from the time we filed this appeal his medical conditions and needs have changed several times. Testimony from his mother will verify this fact. He has difficulty with breathing due to severe sleep apnea requiring careful nighttime monitoring, along with constant infections of the ear, nose and throat. Part of his tongue is so far back in his mouth, a common problem for children with his disorder, that he chokes and requires personnel skilled at medical intervention to assist him, not only when eating, but other times, such as in a swimming pool or even taking a drink of water. He has orthopedic issues and receives regular therapies. His team of specialists' works with his parents and his primary care physician to address [REDACTED]'s substantial needs, often juggling various needs, as he also has fatigue issues and can only tolerate so many medical procedures at one time. Because of his frailty he also cannot be constantly stressed, so testing and sometimes even treatments has to be balanced among health maintenance and managing his frequent infections.

[REDACTED] receives special education services in school, and is cared for at home. His mother [REDACTED] is his primary caregiver. She is not paid to provide care and is not enrolled in a parent C.N.A. program. His father is also involved in his life, provides for him financially, and provides him with medical insurance. Medicaid CHCBS has been providing the extensive medical wrap around services that he has needed and continues to need. It is solely due to the waiver that he has been able to be maintained outside of an institutional setting such as a hospital or skilled nursing facility. His treating physicians and common sense dictate that his loving parents in the community most appropriately care for this seven-year-old child. Without the supports provided by the Medicaid waiver, the family would not be able to afford the extensive therapies, and extraordinary medical care (including frequent diagnostics) necessary to keep him in the community. However, if his mother had to take on a full time job to purchase these services, then [REDACTED] would not have the care he needs at home. [REDACTED] does attend school, but is sent home on a regular basis, due to his frailty and illnesses.

Because of his risk of infection, he cannot attend a typical day care facility. An unskilled person cannot watch him² due to his breathing and choking risks.

██████████ has been a client for years and has received ongoing case management services by Imagine! He receives medical care from a variety of physicians and therapists. There are three components of eligibility for the HCBS waiver and ██████████ meets all three components:

- 1) Financial³: This particular program does not count the income of parents and only counts the income of the child. A child on this program cannot have income that exceeds 300% of the current SSI rate. This amount is \$2022 per month in 2011. ██████████ has no income. Parental assets are also waived (hence the term waiver) for children in this program.
- 2) Functional: Any client receiving long-term care from Colorado Medicaid must pass a functional screen. This screen is called the ULTC 100.2 and is referenced in regulations as being the tool to determine that the client meets the level of care required in a skilled nursing facility.⁴ The regulation references a pediatric component⁵; however, this component has never been developed. When the department in 2003 developed the tool, the intent was to create a pediatric counterpart to address the differences between children and adults. While this was not done, case managers were trained on age appropriate scoring. For example, one would not declare a four year old to be functionally impaired because he or she cannot completely bathe or dress independently. This is because no child at age four can do this task with complete independence. However a typical four year old should be able to be left in a secure play area while a parent is in the next room and should be able to eat a sandwich that is placed in front of the child. This assessment is conducted by case management agencies under contract with the department. There is no dispute in this case that ██████████ meets the functional criteria.
- 3) Target Group Criteria: Each waiver is designed for a specific group of clients. The Children's HCBS waiver is the result of combining two waivers more than a decade ago. One waiver was for children with developmental disabilities and one was for children with physical disabilities. The state must submit these waiver applications to the federal government and in terms of the target criteria, the state checked the box marked medically fragile. The state did not check any other boxes including the boxes for children who are technology dependent, have life threatening conditions or who have developmental disabilities. The waiver has always served all of these children as long as they met the functional screen and what until recently has been the target criteria. The target criterion for this waiver has always been the designation of disabled. Because the children in this waiver are not SSI eligible due to parents' income, they are never determined disabled by the Social Security Administration. Until 2001, the doctor filled out a form called a Med-9. However the federal government informed the state that this was not acceptable and the state then obtained a contractor to assess all children (and some adults for other purposes) using the EXACT standards that the social security administration uses to determine disability.⁶ This makes sense given the state checking the "medically

² 8.525.11(D)1 (these are home health rules that are referenced throughout the HCBS waivers for the purposes of how HCPF differentiates skilled personal care versus unskilled personal care)

³ 8.506.12

⁴ 8.401.1

⁵ 8.506..11 A (4)

⁶ Medicaid letter to counties and case management agencies MA-OI-7-P (attached)

fragile” box because SSA disability is always based on a medical component. In fact, SSA regulations clarify that conditions must be **MEDICALLY DETERMINABLE**.⁷ Social Security generally employs a rigorous evaluation process to determine disability except in those cases where the medical condition is so severe and extreme that it is obvious to any professional in the field that the condition is medically disabling. One of those conditions is Non Mosaic Downs Syndrome⁸, the primary diagnosis held by [REDACTED] Exhibit

There are some other criteria⁹ relevant to this waiver such as the requirement that the child be living at home, in Colorado, and uses a waiver service each month. None of these criteria is in dispute.

⁷ <http://www.socialsecurity.gov/disability/professionals/bluebook/evidentiary.htm>

⁸ This disorder is not listed in the SSA guidelines as a mental or developmental disability but as a disorder affecting multiple body systems: *A. What kinds of impairments do we evaluate under this body system listing?*

1. *General.* We use these listings when you have a single impairment that affects two or more body systems. Under these listings, we evaluate impairments that affect multiple body systems due to non-mosaic Down syndrome or a catastrophic congenital abnormality or disease. These kinds of impairments generally produce long-term, if not lifelong, interference with age-appropriate activities. Some of them result in early death or interfere very seriously with development. We use the term “very seriously” in these listings to describe an “extreme” limitation of functioning as defined in §416.926a(e)(3).

2. *What is Down syndrome?* Down syndrome is a condition in which there are three copies of chromosome 21 within the cells of the body instead of the normal two copies per cell. The three copies may be separate (trisomy), or one chromosome 21 copy may be attached to a different chromosome (translocation). This extra chromosomal material changes the orderly development of the body and brain. Down syndrome is characterized by a complex of physical characteristics, delayed physical development, and mental retardation. Down syndrome exists in non-mosaic and mosaic forms.

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3. *What is non-mosaic Down syndrome?*

a. Non-mosaic Down syndrome occurs when you have an extra copy of chromosome 21 in every cell of your body. At least 98 percent of people with Down syndrome have this form (which includes either trisomy or translocation type chromosomal abnormalities). Virtually all cases of non-mosaic Down syndrome affect the mental, neurological, and skeletal systems, and they are often accompanied by heart disease, impaired vision, hearing problems, and other conditions.

b. We evaluate children with confirmed non-mosaic Down syndrome under 110.06. If you have confirmed non-mosaic Down syndrome, we consider you disabled from birth

⁹ 8.506 et.seq.

The only criterion in dispute is the targeting criteria. In 2007, the federal government agency that has oversight over HCPF did a review of the CHCBS waiver. HCPF failed to meet all six quality assurance standards. The waiver renewal asks HCPF to outline a process for transition if there are areas where they must change something.¹⁰ HCPF responded that they would address transition plans, definitions, and any other transitions in a global quality improvement strategy in 2009.⁹ HCPF developed a strategy but did not share it widely and the strategy did not address the issues in this case and related cases.

Even though HCPF failed all six areas of quality assurance, they focused on one area. HCPF had failed to document that all children being served by the waiver were qualified, generally failing to document that they met either functional or targeting criteria. HCPF at the time went around the state training case managers telling them (and the OAC) that the federal government was requiring that HCPF make a change. Case managers were trained on a new way of assessing clients with new criteria. However, HCPF never promulgated rules, in violation of the Administrative Procedures Act¹¹. Now HCPF is saying that there is not change of policy, but that some case management agencies were not properly implementing existing policy. The reason this is so important is that absent a change in state rule or law, HCPF cannot take Medicaid away from a client who has not had any change in eligibility status. Moreover, the significant federal dollars that HCPF received from the ARRA funds would be at risk if HCPF made a change that created eligibility criteria that would result in people losing services.¹² The response to the waiver HCPF provided to CMS shows that the ULTC 100.2 has always been how waiver targeting criteria was assessed. Please refer to exhibits 2, 3 and 4.

DUE PROCESS ISSUES:

- 1) Was there a change in state rule or law? Per the Colorado Administrative Procedure Act APA, this includes a reinterpretation.
- 2) If there was no such change has there been a change in the child's medical, financial or living situation?
- 3) Are requirements for documentation clear in the regulations and guidance given to the case management agencies?

1) There was a change in how the state interpreted eligibility but no rules or laws were changed. There is no evidence that prior to 2008 or 2009 that there was any requirement for the child to prove medical fragility. Even though that box was checked in the waiver, the state regulations do not define that term. The term is defined in state statute in the human services code at CRS 26-6-102(5.6)¹³.

¹⁰ Waiver application –sub assurances

¹¹ CRS 24-4-103.15

¹² Section 5001 of the American Recovery and Reinvestment Act specifically prohibits states from creating requirements that would make Medicaid eligibility stricter than requirements in place on July 01, 2008.

¹³

5.6) "Medical foster care" means a program of foster care that provides home-based care for medically fragile children and youth who would otherwise be confined to a hospital or institutional setting and includes, but is not limited to, the following: CRS 26.6-102.5.6

This is the only definition in Colorado law or regulation and by any standard; ██████████ is medically fragile using this definition. HCPF also changed their process regarding the Utilization Review Contractor or URC. Until 2009, this was always the case management agency working with the child and doing the functional screen, such as a community centered board, a single entry point or a private case management agency. (See Exhibit X) HCPF regulations at in 8.390.1 T define a URC as follows: Utilization Review Contractor shall mean **an entity or entities** contracted with the Department of Health Care Policy and Financing to provide assessment, case management, training, monitoring, and/or utilization control for the following programs (then lists all HCBS waivers). This means it is an agency with specific skills under contract with HCPF. The waiver application also says that agencies under contract with Medicaid such as Single Entry Point or Community Centered Boards do all level of care assessments.¹⁴ At some point, without a rule change or any transparency Lois Jacobs, a long time state employee and the individual who was responsible for the waiver during the period when HCPF failed to meet all six quality assurance measures, declared herself the URC. This has been going on for more than two years and the state regularly admits that they are aware that their rules are not clear, that there is no pediatric assessment, and continues to change the definition or criteria for the target group. This leads to an environment where decisions about eligibility for a public program are arbitrary and capricious.

(a) Infants impacted by prenatal drug and alcohol abuse;

(b) Children with developmental disabilities which require ongoing medical intervention;

(c) Children and youth diagnosed with acquired immune deficiency syndrome or human immunodeficiency virus;

(d) Children with a failure to thrive or other nutritional disorders; and

e) Children dependent on technology such as respirators, tracheotomy tubes, or ventilators in order to survive.

- a. ¹⁴ **Responsibility for Performing Evaluations and Reevaluations.** Level of care evaluations and reevaluations are performed (*select one*):

Directly by the Medicaid agency

By the operating agency specified in Appendix A

By an entity under contract with the Medicaid agency

If the rule changed, the state has failed to abide by the APA¹⁵ by applying a new standard not in the rules. If this is the case, then HCPF violated their agreement with the federal government under ARRA OR the rules did not change and because there has been no change in Kennedy's medical condition or functional ability, he remains eligible for the program.

Even if an argument can be made that a child must be deemed medically fragile, absent a HCPF definition then a statutory definition that was created for children must be accepted—at least until HCPF comes up with another definition.

HCPF insists that they must comply with terms of the waiver and that somehow the only way to come into compliance is to remove specific children, based on diagnosis rather than medical need in an arbitrary manner. Many areas of the waiver are not current Colorado practice. This includes the fact that the “primary change” the state advertised on page one of the waiver renewal has not been implemented.¹⁶ Moreover, the state has not followed their promise to CMS to have a reasonable transition process and has failed to implement the QIS.¹⁷ This link is for the waiver, which is very long and impossible to print due to the format.

<http://www.cms.gov/MedicaidStWaivProgDemoPGI/MWDL/itemdetail.asp?filterType=dual,%20data&filterValue=Colorado&filterByDID=2&sortByDID=2&sortOrder=ascending&itemID=CMS1217851&intNumPerPage=10>

Finally, if and when HCPF promulgates rules or inserts a new definition into the regulations, that new definition cannot legally be applied until the rule is final and adopted per the APA. For all of these reasons ██████████ remains continually eligible for this program and the denial must be reversed.

Exhibits:

- 1) Resume of Christy Blakely
- 2) Medical information about Downs Syndrome
- 3) Social Security Determination that Downs Syndrome is Medical Condition with High Medical Needs
- 4) Medicaid letter to counties and SEP re Med 9
- 5) CMS letter re failure of all areas of quality
- 6) HCPF explanation to CMS explaining that they did use the ULTC 100.2

¹⁵ CRS 24-4-103(10)

¹⁶ Page 1 of the waiver asks what major changes the state is making, the answer was that the state is implementing the Consumer Directed Attendant Support Services program into this waiver.

¹⁷ Waiver description of how Level of Care is determined: The Case Management Agencies complete a comprehensive assessment utilizing the Universal Long Term Care 100.2 (ULTC 100.2) instrument. The ULTC 100.2 includes a functional assessment and Professional Medical Information Page (PMIP). The functional assessment measures six (6) defined Activities of Daily Living (bathing, eating, mobility, toileting, transferring, or dressing) and/or the need for supervision for behavioral or cognitive dysfunction. The case manager sends the PMIP to the child's medical professional for completion. The medical professional verifies the child's need for institutional level of care. The Department is in the process of developing a child's version of the ULTC 100.2. The targeted implementation date is February, 2009. This will be part of the CHCBS work plan.

- 7) Hilltop Study on Assessment Tools showing ULTC 100.2 is one of the better and more accurate tools showing that people who pass DO require a skilled nursing facility level of care
- 8) MSB Minutes showing that HCPF considers all waiver clients medically fragile (Implementation of SB 04-029 required all medically fragile clients to have an emergency plan, HCPF determined that all waiver clients were by nature medically fragile, minutes and links clarify)

Respectfully submitted,



Julie Reiskin

<http://www.nlm.nih.gov/medlineplus/ency/article/000997.htm>

Down syndrome

Down syndrome is a genetic condition in which a person has 47 chromosomes instead of the usual 46.

Causes

In most cases, Down syndrome occurs when there is an extra copy of chromosome 21. This form of Down syndrome is called Trisomy 21. The extra chromosome causes problems with the way the body and brain develop.

Down syndrome is the most common single cause of human birth defects.

Symptoms

Down syndrome symptoms vary from person to person and can range from mild to severe. However, children with Down syndrome have a widely recognized appearance.

The head may be smaller than normal and abnormally shaped. For example, the head may be round with a flat area on the back. The inner corner of the eyes may be rounded instead of pointed.

Common physical signs include:

- Decreased muscle tone at birth
- Excess skin at the nape of the neck
- Flattened nose
- Separated joints between the bones of the skull (sutures)
- Single crease in the palm of the hand
- Small ears
- Small mouth
- Upward slanting eyes
- Wide, short hands with short fingers
- White spots on the colored part of the eye (Brushfield spots)

Physical development is often slower than normal. Most children with Down syndrome never reach their average adult height.

Children may also have delayed mental and social development. Common problems may include:

- Impulsive behavior
- Poor judgment
- Short attention span
- Slow learning

As children with Down syndrome grow and become aware of their limitations, they may also feel frustration and anger.


Many different medical conditions are seen in people with Down syndrome, including:

- Birth defects involving the heart, such as an atrial septal defect or ventricular septal defect
- Dementia may be seen
- Eye problems, such as cataracts (most children with Down syndrome need glasses)
- Early and massive vomiting, which may be a sign of a gastrointestinal blockage, such as esophageal atresia and duodenal atresia
- Hearing problems, probably caused by regular ear infections
- Hip problems and risk of dislocation
- Long-term (chronic) constipation problems
- Sleep apnea (because the mouth, throat, and airway are narrowed in children with Down syndrome)
- Teeth that appear later than normal and in a location that may cause problems with chewing
- Underactive thyroid (hypothyroidism)



Heart-Related Syndromes

Down Syndrome (Trisomy 21)

Down syndrome, also known as Trisomy 21, is the single most common genetic pattern of malformation in man.  **Glossary**

Most text books quote the incidence of Down syndrome to be between one in 700 to 800 live births.

In 1866, John Langdon Haydon Down described the physical features and associated medical problems that have come to be known as Down syndrome.

In the 1930s, physicians established a relationship between advanced maternal age and Down syndrome. The chromosomal, or genetic, basis of Down syndrome was not established until 1959.

A normal human cell contains 23 pairs of chromosomes which carry all of a person's genetic information. Due to several possible abnormal mechanisms of cell reproduction, patients with Down syndrome have an extra (third) copy of the 21st chromosome. Thus, Down syndrome is also called Trisomy 21.

Advanced maternal age is associated with a high incidence of Trisomy 21, but even women of typical child bearing age can have affected babies.

While the diagnosis may be strongly suggested by characteristic physical findings, the final diagnosis is often made only after chromosome analysis, which includes a complete count and visualization under a microscope of the chromosomes taken from cells in the blood.

A geneticist most often performs this along with a clinical evaluation.

Physical Characteristics of Down Syndrome

The physical characteristics of Down Syndrome in infancy typically involve low muscle tone, flat appearance of the face, upward slanting eye creases, small ears, single skin crease in the palm, extremely flexible joints, large tongue, and several others.

Physical characteristics may be very obvious, but sometimes are so subtle that the diagnosis can only be made after testing is completed. Physical findings do change as the child ages and certain features may become more or less noticeable with time.

Non-cardiac Medical Problems Associated with Down Syndrome

While this chapter focuses on the heart problems associated with Down syndrome, some of the other medical problems seen with Trisomy 21 are:

1. Developmental delay -- All children with Down syndrome are delayed, although this may not be apparent until the child is beyond infancy.
2. Gastrointestinal abnormalities -- 2 percent to 5 percent of children have complete obstruction of the small bowel known as duodenal atresia. Another 2 percent have poor movement abilities of the colon and / or rectum known as Hirschsprung disease.
3. Hearing loss -- Some degree of hearing loss is present in 40 percent to 75 percent of children with Down syndrome. Malformations

Related Services

- Heart Institute
- Thomas Center for Down Syndrome

Clinical Trials / Research Studies

- Children 18-36 Months Old with Down Syndrome Needed for Language Therapy Study
- For Children and Adults of All Ages with Down Syndrome
- A Survey for Parents of Teens With Developmental Disabilities

Down Syndrome Resources

- Cincinnati Children's Down Syndrome Program
- Cincinnati Children's Jack H. Rubinstein Library Down Syndrome Resources
- The Down Syndrome WWW Page
- The National Down Syndrome Society
- National Association for Down Syndrome
- Down Syndrome Association of Greater Cincinnati

may affect either the middle or inner ear structures. Ear infections can be a frequent, often difficult problem to treat.

4. Eye disorders -- 60 percent of children have eye disorders that need monitoring and treatment. Congenital cataracts (loss of transparency of the lens of the eye), glaucoma (increased pressure within the eye), strabismus (cross-eyed) and major refractory errors (far sighted or near sighted) are the common problems.
5. Leukemia -- Leukemia occurs in one of every 150 children with Down Syndrome. This is 20 times higher than the general population.
6. Thyroid disorders -- Low thyroid levels are seen in approximately 5 percent of children with Down syndrome.

Congenital Heart Disease Associated with Down Syndrome

The overall incidence of congenital heart disease in the general population is 0.8 percent. The incidence of congenital heart disease in children with Down syndrome is up to 50 percent.

The types of heart defects in children with Down syndrome can be broken down into three broad categories:

1. Atrioventricular septal defects
2. Ventricular septal defect (VSD), atrial septal defect, or patent ductus arteriosus
3. Other complex heart disease

Atrioventricular septal defects (AV Canal) is greatly over-represented in these children, making up approximately 60 percent of the congenital heart disease found in Trisomy 21. For comparison, AV canals account for only 2.9 percent of the congenital heart defects in the general population.

Ventricular septal defects, atrial septal defects, and patent ductus arteriosus comprise another 20 percent of the congenital heart disease associated with Down syndrome, although these defects are much more common in the general population compared to AV canals.

Other complex congenital heart diseases including Tetralogy of Fallot and hypoplastic left heart syndrome comprise the rest of the heart disease seen in Trisomy 21.

In addition to the heart defects associated with Down Syndrome, isolated elevated blood pressure in the lungs (pulmonary hypertension) is seen with higher frequency in patients with Down syndrome. This high pressure may be related to malformation of the lung tissue, although the exact cause is not known.

The higher pressures may limit the amount of blood flow to the lungs and therefore decrease the likelihood of symptoms of congestive heart failure seen in babies with complete AV canals or large ventricular septal defects.

However, since the pulmonary hypertension can become irreversible, particularly if these large holes are not surgically corrected by a year of age, children with Down syndrome and AV canals or large ventricular septal defects are often referred for surgery earlier than a non-Down syndrome child with the same heart defect.

If the baby does have evidence of high pressures in the lungs, it may complicate postoperative management and hence prolong the recovery time.

Signs and Symptoms of Children with Down Syndrome and Congenital Heart Disease

Depending on the type of heart defect, children with Down syndrome and heart disease may have a variety of symptoms.

Babies with complete AV canal defects may have signs and symptoms of congestive heart failure such as a fast respiratory rate, labored breathing, difficulty completing feedings, sweating with feedings and poor weight gain.

Children with partial AV canals can be asymptomatic because the extra blood flow to the lungs is not enough to cause symptoms of congestive heart failure.

Even babies with large ventricular septal defects as part of their heart disease who should have large amounts of extra blood flowing to their lungs may have no symptoms due to high pressures in the lungs. These babies must be watched carefully to determine the proper timing for

surgery.

Diagnosis of Congenital Heart Disease in Babies with Trisomy 21

Most cardiologists would agree that all babies that have been diagnosed with Down syndrome should have a cardiology evaluation because of the high incidence of associated congenital heart defects.

What degree of cardiac evaluation remains controversial. A good history and physical examination should be performed by an experienced cardiologist to rule out any obvious heart defect.

Most cardiologists would perform an electrocardiogram, which can be very helpful in making the diagnosis of AV canal, even in the absence of physical findings.

Some cardiologists would be satisfied with this level of evaluation. There are those, though, who would argue that at least one echocardiogram should be performed to rule out subtle heart problems.

If the diagnosis of congenital heart disease is made, the cardiologist will follow the child with office visits on a fairly regular basis. Most patients can be watched clinically to determine if the child is having difficulty due to the heart defect.

Occasionally, a repeat electrocardiogram, chest X-ray, or echocardiogram is performed to further evaluate clinical changes. These tests are likely to be repeated before surgical repair is recommended.

Rarely, a cardiac catheterization is required for complete evaluation prior to corrective surgery, especially in patients where elevated pressures in the lungs are a concern.

Treatment Options for Children with Down Syndrome and Congenital Heart Disease

Children with Down syndrome and symptoms of congestive heart failure can be initially managed medically with the use of diuretics, blood pressure medications to allow the heart to eject more blood out to the body rather than out to the lungs, and / or digoxin, a medication used to improve the pumping ability of the heart.

If the baby is having difficulty with feeding and weight gain, nasogastric tube feedings with high calorie formula or fortified breast milk can be used to help with growth.

These are all temporary solutions to allow the baby to grow while deciding if and when surgery is indicated. In general, atrial septal defects, ventricular septal defects, and AV canal defects are closed surgically if the child is demonstrating congestive heart failure that cannot be controlled with medication.

If the baby has no signs of heart failure or is controlled well with medications, the decision for surgical closure can be delayed. AV canals are usually repaired electively between 3 and 6 months of age.

Depending on the size of an atrial septal defect or a ventricular septal defect, surgery can be postponed even longer, keeping in mind the risk of developing irreversible high pressures in the lungs.

The decision must be individualized to each child's physical state as well as the family's concerns.

Long-term Prognosis of Babies with Down Syndrome and Congenital Heart Disease

Overall, survival beyond one year of age is 85 percent in all children with Down syndrome. Over 50 percent of individuals with Down syndrome live to be greater than 50 years old.

Congenital heart disease is the most common cause of death in early childhood. However, as of the late 1980s, 70 percent of children with Down syndrome and congenital heart disease lived beyond their first birthday. With improved medical and surgical care, these numbers continue to improve.

Last Updated: 08/2009



As a not-for-profit hospital and research center, Cincinnati Children's Hospital Medical Center is dedicated to bringing the world the joy of healthier kids.

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DOWN SYNDROME HEALTH CARE GUIDELINES

(Based on 1999 Down Syndrome Health Care Guidelines)*

Childhood (1-12 Years)

- < TSH and T4-Thyroid Function Test (annual).
- < Echocardiogram by a pediatric cardiologist if not done previously.
- < Behavioral Auditory Testing (every 6 months to age 3, then annually).
- < Lateral cervical spine x-rays (neutral view, flexion, extension) to rule out atlanto-axial instability. Radiologist to measure atlanto-dens distance and neural canal width (at 3-5 years, then as needed).
- < Pediatric and neurological exam with evaluation for spinal cord compression: deep tendon reflexes, gait, Babinski sign.
- < Use Down syndrome growth charts and head circumference charts with others.
- < Eye examination (annually, or as indicated).
- < Screen for celiac disease IgA antiendomysium antibodies and total IgA (btwn 2 & 3 yrs)
- < Question about obstructive sleep apnea; ENT exam (as needed).
- < Dental Exam (2 years; follow up exams every 6 months after). Twice daily brushing
- < Reinforce need for subacute bacterial endocarditis prophylaxis (SBE) for cardiac problems (as indicated).
- < Brief vulvar exam for girls.
- < Well child care: immunizations; pneumococcal vaccine (2 years).
- < Evaluation by speech/language pathologist.



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*[HEALTH CARE GUIDELINES FOR INDIVIDUALS WITH DOWN SYNDROME: 1999 REVISION (Down Syndrome Preventive Medical Check List) is published in **Down Syndrome Quarterly** (Volume 4, Number 3, September, 1999, pp. 1-16) and is reprinted, duplicated, and/or transmitted with permission of the Editor. Information concerning publication policy or

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Listing of Impairments - Childhood Listings (Part B)

The following sections provide medical criteria for the evaluation of impairments of children under age 18 (where criteria in Part A - Adult listings do not give appropriate consideration to the particular disease process in childhood).

This electronic version contains the new Special Senses and Speech Listings effective August 2, 2010.

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110.00 IMPAIRMENTS THAT AFFECT MULTIPLE BODY SYSTEMS

<u>Section</u>	<u>110.00 IMPAIRMENTS THAT AFFECT MULTIPLE BODY SYSTEMS</u>
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110.01
Category of
Impairments,
Impairments That
Affect Multiple
Body Systems

110.06
Non-mosaic Down
syndrome

110.08
A catastrophic
congenital
abnormality or
disease

A. *What kinds of impairments do we evaluate under this body system listing?*

1. *General.* We use these listings when you have a single impairment that affects two or more body systems. Under these listings, we evaluate impairments that affect multiple body systems due to non-mosaic Down syndrome or a catastrophic congenital abnormality or disease. These kinds of impairments generally produce long-term, if not lifelong, interference with age-appropriate activities. Some of them result in early death or interfere very seriously with development. We use the term “very seriously” in these listings to describe an “extreme” limitation of functioning as defined in §416.926a(e)(3).

2. *What is Down syndrome?* Down syndrome is a condition in which there are three copies of chromosome 21 within the cells of the body instead of the normal two copies per cell. The three copies may be separate (trisomy), or one chromosome 21 copy may be attached to a different chromosome (translocation). This extra chromosomal material changes the orderly development of the body and brain. Down syndrome is characterized by a complex of physical characteristics, delayed physical development, and mental retardation. Down syndrome exists in non-mosaic and mosaic forms.

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3. *What is non-mosaic Down syndrome?*

a. Non-mosaic Down syndrome occurs when you have an extra copy of chromosome 21 in every cell of your body. At least 98 percent of people with Down syndrome have this form (which includes either trisomy or translocation type chromosomal abnormalities). Virtually all cases of non-mosaic Down syndrome affect the mental, neurological, and skeletal systems, and they are often accompanied by heart disease, impaired vision, hearing problems, and other conditions.

b. We evaluate children with confirmed non-mosaic Down syndrome under 110.06. If you have confirmed non-mosaic Down syndrome, we consider you disabled from birth.

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4. *What is mosaic Down syndrome?*

a. Mosaic Down syndrome occurs when you have some cells with the normal two copies of chromosome 21 and some cells with an extra copy of chromosome 21. When this occurs, there is a mixture of two types of cells.

Mosaic Down syndrome occurs in only 1-2 percent of people with Down syndrome, and there is a wide range in the level of severity of the impairment. Mosaic Down syndrome can be profound and disabling, but it can also be so slight as to be undetected clinically.

b. We evaluate children with confirmed mosaic Down syndrome under the listing criteria in any affected body system(s) on an individual case basis, as described in 110.00C.

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5. *What are catastrophic congenital abnormalities or diseases?*

a. Catastrophic congenital abnormalities or diseases are present at birth, although they may not be apparent immediately. They cause deviation from, or interruption of, the normal function of the body and are reasonably certain to result in early death or to interfere very seriously with development.

b. We evaluate catastrophic congenital abnormalities or diseases under 110.08.

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B. *What documentation do we need to establish that you have an impairment that affects multiple body systems?*

1. *General.* We need documentation from an acceptable medical source, as defined in §§404.1513(a) and 416.913(a), to establish that you have a medically determinable impairment. In general, the documentation should include a clinical description of the diagnostic physical features associated with your multiple body system impairment, and any appropriate laboratory tests.

2. *Non-mosaic Down syndrome (110.06).*

a. *Definitive chromosomal analysis.* We will find that you have non-mosaic Down syndrome based on a report from an acceptable medical source that indicates that you have the impairment and that includes the actual laboratory report of definitive chromosomal analysis showing that you have the impairment. Definitive chromosomal analysis for Down syndrome means karyotype analysis. When we have the laboratory report of the actual karyotype analysis, we do not additionally require a clinical description of the physical features of Down syndrome.

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b. *What if you have Down syndrome and we do not have*

the results of definitive chromosomal analysis? When you have Down syndrome and we do not have the actual laboratory report of definitive chromosomal analysis, we need evidence from an acceptable medical source that includes a clinical description of the diagnostic physical features of your impairment, and that is persuasive that a positive diagnosis has been confirmed by definitive chromosomal analysis at some time prior to our evaluation. To be persuasive, the report must state that definitive chromosomal analysis was conducted and that the results confirmed the diagnosis. The report must be consistent with other evidence in your case record; for example, evidence showing your limitations in adaptive functioning or signs of a mental disorder that can be associated with non-mosaic Down syndrome, your educational history, or the results of psychological testing.

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3. Catastrophic congenital abnormalities or diseases (110.08).

a. *Genetic disorders.* For genetic multiple body system impairments (other than non-mosaic Down syndrome), such as Trisomy 13 (Patau Syndrome or Trisomy D), Trisomy 18 (Edwards' Syndrome or Trisomy E), chromosomal deletion syndromes (for example, deletion 5p syndrome, also called cri du chat syndrome), or inborn metabolic disorders (for example, Tay-Sachs disease), we need evidence from an acceptable medical source that includes a clinical description of the diagnostic physical features of your impairment, and the report of the definitive laboratory study (for example, genetic analysis or evidence of biochemical abnormalities) that is diagnostic of your impairment. When we do not have the actual laboratory report, we need evidence from an acceptable medical source that is persuasive that a positive diagnosis was confirmed by appropriate laboratory analysis at some time prior to our evaluation. To be persuasive, the report must state that the appropriate definitive laboratory study was conducted and that the results confirmed the diagnosis. The report must be consistent with other evidence in your case record.

b. *Other disorders.* For infants born with other kinds of catastrophic congenital abnormalities (for example, anencephaly, cyclopia), we need evidence from an acceptable medical source that includes a clinical description of the diagnostic physical features of the impairment.

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C. How do we evaluate impairments that affect multiple body systems and that do not meet the criteria of the listings in this body system?

1. These listings are examples of impairments that commonly affect multiple body systems and that we consider significant enough to result in marked and severe functional limitations. If your severe impairment(s) does not meet the criteria of any of these listings, we must also consider whether your impairment(s) meets the criteria of a listing in another body system.

2. There are many other impairments that can cause deviation from, or interruption of, the normal function of the body or interfere with development; for example, congenital anomalies, chromosomal disorders, dysmorphic syndromes, metabolic disorders, and perinatal infectious diseases. In these impairments, the degree of deviation or interruption may vary widely from child to child. Therefore, the resulting functional limitations and the progression of those limitations are more variable than with the catastrophic congenital abnormalities and diseases we include in these listings. For this reason, we evaluate the specific effects of these impairments on you under the listing criteria in any affected body system(s) on an individual case basis. Examples of such impairments include, but are not limited to, triple X syndrome (XXX syndrome), fragile X syndrome, phenylketonuria (PKU), caudal regression syndrome, and fetal alcohol syndrome.

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3. If you have a severe medically determinable impairment(s) that does not meet a listing, we will consider whether your impairment(s) medically equals a listing. If your impairment(s) does not meet or medically equal a listing, we will consider whether it functionally equals the listings. (See §§404.1526, 416.926, and 416.926a.) When we decide whether you continue to be disabled, we use the rules in §416.994a.

110.01 Category of Impairments, Impairments That Affect Multiple Body Systems

110.06 *Non-mosaic Down syndrome*, established as described in 110.00B.

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110.08 *A catastrophic congenital abnormality or disease*, established as described in 110.00B, and:

A. Death usually is expected within the first months of life, and the rare individuals who survive longer are profoundly

impaired (for example, anencephaly, trisomy 13 or 18, cyclopia);

Or

B. That interferes very seriously with development; for example, cri du chat syndrome (deletion 5p syndrome) or Tay-Sachs disease (acute infantile form).

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