CD Part 10

Multiple Body System Disorders

A. Definitions

The following definitions are for words used in this chapter and during the SSA disability process. If you need additional definitions, consult a good medical dictionary, available in most bookstores and libraries. You can also look at online medical dictionaries like the one at www.medlineplus.gov.

Acquired disorders. Disorders that are not of genetic (hereditary) origin.

Age-appropriate activities. Activities normally expected for a child's age.

Anencephaly. Birth defect characterized by absence of most of the brain.

Anus. Terminal opening of the gastrointestinal tract, through which feces exit the body.

Brushfield spots. White speckles in the iris of the eyes of those with *Down syndrome*.

Cerebrospinal fluid (CSF). Clear, watery, fluid that bathes the spinal cord and brain and circulates within the brain.

Chromosomes. The structures in the nuclei of cells that carry the genetic (DNA) content of an individual. Cells normally have 46 chromosomes, counting the sex chromosomes—22 pairs of somatic chromosomes and one pair of sex chromosomes. The sex chromosomes in males are one X and one Y chromosome (XY) and in females, two X chromosomes (XX). The gene make-up of a normal female would be written 46, XX, and a normal male would be 46, XY.

Congenital. Dating from the time of birth. Congenital disorders are not necessarily hereditary, because some congenital disorders arise from events that happen while a fetus is in the uterus rather than as a result of abnormal genes.

Cyclopia. Birth defect characterized by having only one centrally placed eye and other abnormalities.

Deoxyribonucleic acid (DNA). The chemical structure in *chromosomes* of which *genes* are composed.

Down syndrome. Hereditary disorder involving the presence of an extra #21 *chromosome* in cells, associated with mental retardation and possible abnormalities in various organs such as the heart. Also known as Trisomy 21.

Epicanthic folds. Folds of tissue over the inner corners of the eyes.

Extrauterine life. Life after birth.

Facies. The appearance of the face. In some physical disorders, such as Down syndrome and fetal alcohol syndrome, there is a characteristic facies related to the physical appearance of the face. When discussing mental disorders, facies may be used to describe emotional expression, such as "The depressed child had a sad facies."

Fetal alcohol syndrome. Abnormalities in a child resulting from the mother using alcohol during pregnancy.

Fragile X syndrome. A common hereditary disorder associated with breakage in the X chromosome and characterized by mental retardation as well as other possible abnormalities.

Genes. Pieces of DNA that make up control units for the growth and function of an organism.

Hereditary disorders. Disorders resulting from genetic abnormalities.

Herpes encephalitis. Infection of the brain with herpes virus.

Hydrocephalus. Accumulation of excessive cerebrospinal fluid in the ventricles of the brain. Hydrocephalus may be associated with increased pressure in the ventricles or with normal pressures. The latter is known as normal pressure hydrocephalus (NPH).

Hypothyroidism. Disorder in which there is a deficiency in thyroid hormone. Also known as *myxedema*.

Hypotonia. Poor muscle tone. A muscle with no tone is called flaccid.

Imperforate anus. Absence of an *anus*.

Intestinal atresia. Failure of the intestine to develop.

Macrocephaly. An abnormally large head, such as may be seen with *hydrocephalus*.

Microcephaly. Abnormally small head, such as may be seen with some genetic disorders associated with severe mental retardation.

Mosaic. Any genetic disorder in which different cells in the body have different genetic make-ups. For example, if some cells have an extra chromosome #21 and some cells have a normal number of chromosomes, then the person is said to be a mosaic for *Down syndrome*.

Motor dysfunction. Any abnormality related to movement, such as weakness, paralysis, tremors, or lack of coordination.

Motor function. Abilities related to movement, such as walking and use of the hands and arms.

Multiple body system disorders. Disorders that affect more than one type of organ, such as the heart and brain.

Phenylketonuria (PKU). Hereditary enzyme deficiency associated with mental retardation.

Postural reaction deficit. Decreased ability to recover balance when disturbed.

Primitive reflexes. Reflexes seen in newborn infants, such as rooting and sucking reflexes.

Protozoan. Microscopic animal.

Syndrome. A set of signs and symptoms that occur together.

Tay-Sachs disease. Hereditary enzyme deficiency resulting in severe mental retardation.

Ventricles (of brain). Any one of the connected cavities in the brain that contain *cerebrospinal fluid*.

B. General Information

Down syndrome and other genetic disorders must be diagnosed with chromosomal analysis as well as physical examination. However, the SSA does not require actual laboratory reports if the available medical records convincingly state that the appropriate test confirming the diagnosis was done at some time in the past. For example, a treating doctor might refer in medical records to chromosomal analysis confirming Down syndrome in a child at some time in the past. If the records clearly show the physical abnormalities of Down syndrome, the SSA would not require the claimant to undergo repeat chromosomal testing.

Regarding children, disorders that qualify under these listings are those associated with life-threatening catastrophic congenital abnormalities and other serious hereditary, congenital, or acquired disorders that affect two or more body systems and are expected to:

1. Result in early death or attainment of a developmental level less than that expected of a two-year-old child, such as anencephaly or Tay-Sachs disease (Listing 110.08).
2. Produce significant and long-term—if not lifelong—interference with the child's ability to carry on age-appropriate activities, defined as activities that most other children of the same age can do. The SSA considers such significant interference to exist when a child is not capable of abilities possessed by a normal child two-thirds or less of their age and when the lag in development has lasted, or could be expected to last, at least 12 months. For example, suppose a nine-year-old child claimant is given a test of developmental abilities and gets a score indicating overall abilities (motor abilities, eating, language, etc.) of a six-year-old child. The child's developmental age would be six years, or two-thirds that of a normal nine-year-old. (Listing 110.06.)

Regarding adults, Listing 10.06 is the same as child Listing 110.06.

Chromosomal abnormalities other than full-blown Down syndrome—for example, mosaic Down syndrome, fragile X syndrome, phenylketonuria, and fetal alcohol syndrome, produce a pattern of multiple impairments with a wide range of severity. Therefore, the effects of these impairments should be evaluated under whatever listings deal with the actual disorder. For instance, heart disease would be evaluated under the appropriate heart disease listing—such as the listing for heart failure, if that is the problem (CD Part 4).

Because of the possible involvement of multiple body systems—such as the heart and brain—the SSA should always consider the combined effect of multiple impairments. The SSA must decide whether they are equal in severity to a combination of listings, even if the child or adult has no single impairment that would satisfy a listing. As in the evaluation of any impairment, these determinations should be made by a doctor—not an examiner, claim manager, or other layperson.

C. Specific Listings and Residual Functional Capacity

The listings that follow are in the federal regulations. They have been interpreted and commented on for greater ease of understanding while explaining their requirements. It is impossible to discuss here all of

the medical possibilities related to every kind of disorder, and you may need to seek help from your treating doctor to more fully understand how your particular impairment relates to these listings. The discussion of residual functional capacity does not apply to children.

1. Listing 10.06: Non-Mosaic Down Syndrome (Adults)

Down syndrome (trisomy 21) is a genetic disorder involving the presence of three copies of chromosome #21 in cells, instead of the normal two copies. The presence of this extra chromosome always leads to mental retardation. There is a characteristic facies: flattened bridge of the nose, malformed ears, a large protruding tongue, flattening of the back of the skull, Brushfield spots, epicanthic folds, and a slanting of the eyes. Other features of Down syndrome are malformed hands with short fingers and a wide palm with a prominent crease ("simian crease"). Down syndrome is the most common chromosomal abnormality producing a syndrome in humans. The chances of having a Down syndrome baby steadily increase with the age of the mother. In addition to the above characteristic abnormalities, Down syndrome may be associated with congenital heart disease and intestinal malformations, such as imperforate anus and intestinal atresia.

Only a valid diagnosis of non-mosaic Down syndrome is required. Then the required severity as described above is automatically considered to exist. (See "General Information," description 2, above.)

This listing applies only to claimants who have full-blown (non-mosaic) Down syndrome, in which all of the body's cells carry the abnormal extra #21 chromosome. About 95–99% of Down syndrome cases are non-mosaic and so would qualify.

Cases of mosaic Down syndrome, in which some of the body's cells are normal and others abnormal, are not evaluated under this listing. Mosaics vary in the severity of their disorder; some may have normal intelligence and very little evidence of any significant physical abnormality. Those unusual claimants with mosaic Down syndrome would be evaluated under whatever listings deal with the physical or mental disorders they have associated with their Down syndrome, such as described above. For instance, heart disease would be evaluated under the cardiovascular disease listings (CD Part 4) and sub-average intelligence would be evaluated under the mental disorders listings (CD Part 12).

a. Listing Level Severity

For the applicant's condition to be severe enough to meet the listing, the applicant must have nonmosaic Down syndrome. A chromosomal analysis alone is enough to satisfy the listing, provided that the SSA can obtain an actual copy of the laboratory report. The chromosomal analysis must be clearly diagnostic and be what is known as a *karyotype* chromosomal analysis. In other words, the analysis must include evaluation of the number and shape of chromosomes.

If a chromosomal analysis report is not available, satisfying the listing requires much more information. A detailed physical examination showing the characteristic features of Down syndrome must be provided to the SSA. Also, the SSA will require a statement from the treating doctor that a chromosomal analysis was done at some time in the past and confirmed the diagnosis of Down syndrome. Finally, this report must not conflict with other evidence the SSA may collect—such as educational history, psychological test results, or level of personal functioning.

b. Residual Functional Capacity

An analysis of RFC is not necessary under this listing, since diagnosis of nonmosaic Down syndrome is an automatic allowance. RFC for mosaic Down syndrome would depend on the types of problems present, if any, and reference should be made to the RFC discussion under the appropriate listings for the types of disorders involved.

2. Listing 110.06: Non-Mosaic Down Syndrome (Children)

See comments under adult Listing 10.06. As in adults, full-blown Down syndrome is an automatic allowance.

a. Listing Level Severity

For the child's condition to be severe enough to meet this listing, the child must have non-mosaic Down

syndrome, established by physical examination and chromosomal analysis. The SSA will consider the child disabled from birth.

3. Listing 110.08: Catastrophic Congenital Abnormalities (Children)

Catastrophic abnormalities at the time of birth are those so severe that only brief survival is possible or growth and development will predictably stop at such a young age that the child will always remain helpless as a result of severe mental retardation.

a. Listing Level Severity

For the child's condition to be severe enough to meet this listing, the child must have catastrophic congenital abnormalities that satisfy part Ⓐ or Ⓑ, below.

Ⓐ Part Ⓐ concerns those genetic disorders that are not usually compatible with survival after birth. Examples include anencephaly, cyclopia (one eye, brain malformation, and a fleshy protuberance on the forehead), and other lethal abnormalities such as trisomy D or E. Trisomy D is an older term for trisomy-13, in which there is an extra chromosome #13 in body cells instead of the normal two. Trisomy E is an older term for trisomy-18, in which there is an extra chromosome #18 in body cells instead of the normal two. In both of these disorders, infants have mental retardation, heart disease, kidney disease, and other abnormalities. Trisomy 18 is common, being the second most frequent abnormality of nonsex chromosomes in humans and occurring in about one in 8,000 births. In the above examples, death occurs within several months of birth, although there may be very rare exceptions.

Ⓑ A positive diagnosis where the child is not expected to ever attain the growth and developmental level of a two-year-old. Examples include the genetic disorders of Tay-Sachs disease and cri du chat syndrome, but allowance is not limited to those disorders.

In Tay-Sachs disease there is deficiency of an enzyme leading to the accumulation of compounds that damage the brain. Severe degeneration of the brain is evident on imaging studies of that organ. Development may progress normally until about five months of age, but there follows severe mental retardation, blindness, and other serious abnormalities such as hypotonia and seizures. There is also a late-onset form of Tay-Sachs that may not be seen until adulthood.

Cri du chat syndrome is caused by a missing piece of chromosome #5. These children are mentally retarded, with microcephaly and malformed heads, skeletal abnormalities, and possibly congenital heart disease. ■