



SOCIAL SECURITY ADMINISTRATION

20 CFR Part 404

[Docket No. SSA-2009-0039]

RIN 0960-AH04

Revised Medical Criteria for Evaluating Congenital Disorders That Affect Multiple Body Systems

AGENCY: Social Security Administration.

ACTION: Final rule.

SUMMARY: We are revising the criteria in the Listing of Impairments (listings) that we use to evaluate cases involving impairments that affect multiple body systems in adults and children under titles II and XVI of the Social Security Act (Act). The revisions reflect our program experience and address adjudicator questions we have received since we last comprehensively revised this body system in 2005. We do not expect any decisional differences due to the revisions in this body system.

DATES: These rules are effective [INSERT DATE 60 DAYS AFTER DATE OF PUBLICATION IN THE FEDERAL REGISTER].

FOR FURTHER INFORMATION CONTACT: Cheryl Williams, Office of Medical Listings Improvement, Social Security Administration, 6401 Security Boulevard,

Baltimore, Maryland 21235-6401, (410) 965-1020. For information on eligibility or filing for benefits, call our national toll-free number, 1-800-772-1213, or TTY 1-800-325-0778, or visit our Internet site, Social Security Online, at <http://www.socialsecurity.gov>.

SUPPLEMENTARY INFORMATION:

Background

We are making final the rules for evaluating congenital disorders that affect multiple body systems we proposed in a notice of proposed rulemaking (NPRM) we published in the Federal Register on October 25, 2011 (76 FR 66006). The preamble to the NPRM provides a full explanation of the background of these revisions. We are not repeating that information here because we are adopting our proposed rules without change. You can view the preamble to the NPRM by visiting www.regulations.gov and searching for document “SSA-2009-0039-0004.”

Why are we revising the listings for evaluating congenital disorders that affect multiple body systems?

We are revising the listings for evaluating congenital disorders that affect multiple body systems to update the medical criteria, clarify how we evaluate congenital disorders, and address adjudicator questions.

When will we begin to use these final rules?

We will begin to use these final rules on their effective date. We will continue to use the current listings until the date these final rules become effective. We will apply the final rules to new applications filed on or after the effective date of these final rules and to claims that are pending on or after the effective date.¹ These final rules will remain in effect for 5 years after the date they become effective, unless we extend them, or revise and issue them again.

Public Comments

In the NPRM, we provided the public with a 60-day comment period, which ended on December 27, 2011. We received one public comment letter. The comment came from a national group representing disability examiners in the State agencies that make disability determinations for us.

¹ This means that we will use these final rules on and after their effective date, in any case in which we make a determination or decision. We expect that Federal courts will review our final decisions using the rules that were in effect at the time we issued the decisions. If a court reverses the our final decision and remands a case for further administrative proceedings after the effective date of these final rules, we will apply these final rules to the entire period at issue in the decision we make after the court's remand.

Below we provide a summary of points that were relevant to this rulemaking and our responses. We tried to present the commenter's concerns and suggestions accurately and completely.

Comment: The commenter suggested revisions to the proposed criteria for meeting listings 10.06 and 110.06 Non-mosaic Down syndrome. The commenter suggested that an individual be found to meet the criteria of the listings unless chromosomal analysis shows a diagnosis of mosaic Down syndrome.

Response: We are not adopting this comment because we do not agree with the suggestion that an individual should be found to meet listings 10.06 or 110.06 unless chromosomal analysis shows a diagnosis of mosaic Down syndrome. We believe that the evidence needs to confirm a diagnosis of non-mosaic Down syndrome. Our rules specify that mosaic Down syndrome does not meet the criteria of our listings. However, it could satisfy the criteria of listings in other body systems, depending on the severity of the manifestations.

Comment: The commenter also stated that fluorescence in situ hybridization (FISH) testing could differentiate non-mosaic from mosaic Down syndrome. The commenter suggested that we use this test in combination with a clinical description of diagnostic physical features and a diagnosis from an acceptable medical source to meet listings 10.06 and 110.06.

Response: We do not agree that we should use FISH testing when we evaluate non-Mosaic Down syndrome under listings 10.06 and 110.06. FISH testing does not distinguish between mosaic and non-mosaic Down syndrome. Karyotype analysis is the only stand-alone method of chromosomal analysis acceptable for confirming non-mosaic Down syndrome.

What is our authority to make rules and set procedures for determining whether a person is disabled under the statutory definition?

The Act authorizes us to make rules and regulations and to establish necessary and appropriate procedures to implement them. Sections 205(a), 702(a)(5), and 1631(d)(1).

Regulatory Procedures

Executive Order 12866, as Supplemented by Executive Order 13563

We have consulted with the Office of Management and Budget (OMB) and determined that these final rules meet the criteria for a significant regulatory action under Executive Order 12866, as supplemented by Executive Order 13563. Therefore, OMB reviewed them.

Regulatory Flexibility Act

We certify that these final rules will not have a significant economic impact on a substantial number of small entities because they affect individuals only. Therefore, the Regulatory Flexibility Act, as amended, does not require us to prepare a regulatory flexibility analysis.

Paperwork Reduction Act

These rules do not create any new or affect any existing collections and, therefore, do not require Office of Management and Budget approval under the Paperwork Reduction Act.

(Catalog of Federal Domestic Assistance Program Nos. 96.001, Social Security – Disability Insurance; 96.002, Social Security – Retirement Insurance; 96.004, Social Security – Survivors Insurance; and 96.006, Supplemental Security Income)

List of Subjects in 20 CFR Part 404

Administrative practice and procedure; Blind, Disability benefits; Old-age, Survivors, and Disability Insurance; Reporting and recordkeeping requirements; Social Security.

Dated:

Michael J. Astrue,
Commissioner of Social Security.

For the reasons set out in the preamble, we are amending 20 CFR part 404 subpart P as set forth below:

PART 404—FEDERAL OLD-AGE, SURVIVORS AND DISABILITY INSURANCE
(1950-)

Subpart P—[Amended]

1. The authority citation for subpart P of part 404 continues to read as follows:

Authority: Secs. 202, 205(a)-(b) and (d)-(h), 216(i), 221(a), (i), and (j), 222(c), 223, 225, and 702(a)(5) of the Social Security Act (42 U.S.C. 402, 405(a)-(b) and (d)-(h), 416(i), 421(a), (i), and (j), 422(c), 423, 425, and 902(a)(5)); sec. 211(b), Pub. L. 104-193, 110 Stat. 2105, 2189; sec. 202, Pub. L. 108-203, 118 Stat. 509 (42 U.S.C. 902 note).

2. Amend appendix 1 to subpart P of part 404 by

- a. Revising item 11 of the introductory text;
 - b. Revising the body system name in part A for section 10.00 in the table of contents;
 - c. Revising section 10.00 in part A;
 - d. Revising the body system name in part B for section 110.00 in the table of contents;
- and
- e. Revising section 110.00 in part B.

The revisions read as follows:

APPENDIX 1 TO SUBPART P OF PART 404—LISTING OF IMPAIRMENTS

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11. Congenital Disorders That Affect Multiple Body Systems (10.00 and 110.00):

[Insert date 5 years from the effective date of the final rules].

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10.00 Congenital Disorders That Affect Multiple Body Systems.

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Part A

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10.00 CONGENITAL DISORDERS THAT AFFECT MULTIPLE BODY SYSTEMS

A. Which disorder do we evaluate under this body system? Although Down syndrome exists in non-mosaic and mosaic forms, we evaluate only non-mosaic Down syndrome under this body system.

B. What is non-mosaic Down syndrome? Non-mosaic Down syndrome is a genetic disorder. Most people with non-mosaic Down syndrome have three copies of chromosome 21 in all of their cells (chromosome 21 trisomy); some have an extra copy of chromosome 21 attached to a different chromosome in all of their cells (chromosome 21 translocation). Virtually all people with non-mosaic Down syndrome have

characteristic facial or other physical features, delayed physical development, and intellectual disability. People with non-mosaic Down syndrome may also have congenital heart disease, impaired vision, hearing problems, and other disorders. We evaluate non-mosaic Down syndrome under 10.06. If you have non-mosaic Down syndrome documented as described in 10.00C, we consider you disabled from birth.

C. What evidence do we need to document non-mosaic Down syndrome under 10.06?

1. Under 10.06A, we will find you disabled based on laboratory findings.

a. To find that your disorder meets 10.06A, we need a copy of the laboratory report of karyotype analysis, which is the definitive test to establish non-mosaic Down syndrome. We will not purchase karyotype analysis. We will not accept a fluorescence in situ hybridization (FISH) test because it does not distinguish between the mosaic and non-mosaic forms of Down syndrome.

b. If a physician (see §§404.1513(a)(1) and 416.913(a)(1) of this chapter) has not signed the laboratory report of karyotype analysis, the evidence must also include a physician's statement that you have Down syndrome.

c. For purposes of 10.06A, we do not require additional evidence stating that you

have the distinctive facial or other physical features of Down syndrome.

2. If we do not have a laboratory report of karyotype analysis showing that you have non-mosaic Down syndrome, we may find you disabled under 10.06B or 10.06C.

a. Under 10.06B, we need a physician's report stating: (i) your karyotype diagnosis or evidence that documents your type of Down syndrome is consistent with prior karyotype analysis (for example, reference to a diagnosis of "trisomy 21"), and (ii) that you have the distinctive facial or other physical features of Down syndrome. We do not require a detailed description of the facial or other physical features of the disorder. However, we will not find that your disorder meets 10.06B if we have evidence—such as evidence of functioning inconsistent with the diagnosis—that indicates that you do not have non-mosaic Down syndrome.

b. If we do not have evidence of prior karyotype analysis (you did not have testing, or you had testing but we do not have information from a physician about the test results), we will find that your disorder meets 10.06C if we have: (i) a physician's report stating that you have the distinctive facial or other physical features of Down syndrome, and (ii) evidence that your functioning is consistent with a diagnosis of non-mosaic Down syndrome. This evidence may include medical or nonmedical information about your physical and mental abilities, including information about your education, work history, or the results of psychological testing. However, we will not find that your

disorder meets 10.06C if we have evidence—such as evidence of functioning inconsistent with the diagnosis—that indicates that you do not have non-mosaic Down syndrome.

D. How do we evaluate mosaic Down syndrome and other congenital disorders that affect multiple body systems?

1. Mosaic Down syndrome. Approximately 2 percent of people with Down syndrome have the mosaic form. In mosaic Down syndrome, there are some cells with an extra copy of chromosome 21 and other cells with the normal two copies of chromosome 21. Mosaic Down syndrome can be so slight as to be undetected clinically, but it can also be profound and disabling, affecting various body systems.

2. Other congenital disorders that affect multiple body systems. Other congenital disorders, such as congenital anomalies, chromosomal disorders, dysmorphic syndromes, inborn metabolic syndromes, and perinatal infectious diseases, can cause deviation from, or interruption of, the normal function of the body or can interfere with development. Examples of these disorders include both the juvenile and late-onset forms of Tay-Sachs disease, trisomy X syndrome (XXX syndrome), fragile X syndrome, phenylketonuria (PKU), caudal regression syndrome, and fetal alcohol syndrome. For these disorders and other disorders like them, the degree of deviation, interruption, or interference, as well as the resulting functional limitations and their progression, may vary widely from person to person and may affect different body systems.

3. Evaluating the effects of mosaic Down syndrome or another congenital disorder under the listings. When the effects of mosaic Down syndrome or another congenital disorder that affects multiple body systems are sufficiently severe we evaluate the disorder under the appropriate affected body system(s), such as musculoskeletal, special senses and speech, neurological, or mental disorders. Otherwise, we evaluate the specific functional limitations that result from the disorder under our other rules described in 10.00E.

E. What if your disorder does not meet a listing? 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. See §§404.1526 and 416.926 of this chapter. If your impairment(s) does not meet or medically equal a listing, you may or may not have the residual functional capacity to engage in substantial gainful activity. We proceed to the fourth, and if necessary, the fifth steps of the sequential evaluation process in §§ 404.1520 and 416.920 of this chapter. We use the rules in §§ 404.1594 and 416.994 of this chapter, as appropriate, when we decide whether you continue to be disabled.

10.01 Category of Impairments, Congenital Disorders That Affect Multiple Body Systems

10.06 Non-mosaic Down syndrome (chromosome 21 trisomy or chromosome 21

translocation), documented by:

A. A laboratory report of karyotype analysis signed by a physician, or both a laboratory report of karyotype analysis not signed by a physician and a statement by a physician that you have Down syndrome (see 10.00C1).

OR

B. A physician's report stating that you have chromosome 21 trisomy or chromosome 21 translocation consistent with prior karyotype analysis with the distinctive facial or other physical features of Down syndrome (see 10.00C2a).

OR

C. A physician's report stating that you have Down syndrome with the distinctive facial or other physical features and evidence demonstrating that you function at a level consistent with non-mosaic Down syndrome (see 10.00C2b).

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110.00 Congenital Disorders That Affect Multiple Body Systems

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Part B

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110.00 CONGENITAL DISORDERS THAT AFFECT MULTIPLE BODY SYSTEMS

A. Which disorders do we evaluate under this body system? We evaluate non-mosaic Down syndrome and catastrophic congenital disorders under this body system.

B. What is non-mosaic Down syndrome? Non-mosaic Down syndrome is a genetic disorder. Most children with non-mosaic Down syndrome have three copies of chromosome 21 in all of their cells (chromosome 21 trisomy); some have an extra copy of chromosome 21 attached to a different chromosome in all of their cells (chromosome 21 translocation). Virtually all children with non-mosaic Down syndrome have characteristic facial or other physical features, delayed physical development, and intellectual disability. Children with non-mosaic Down syndrome may also have congenital heart disease, impaired vision, hearing problems, and other disorders. We evaluate non-mosaic Down syndrome under 110.06. If you have non-mosaic Down syndrome documented as described in 110.00C, we consider you disabled from birth.

C. What evidence do we need to document non-mosaic Down syndrome under 110.06?

1. Under 110.06A, we will find you disabled based on laboratory findings.

a. To find that your disorder meets 110.06A, we need a copy of the laboratory report of karyotype analysis, which is the definitive test to establish non-mosaic Down syndrome. We will not purchase karyotype analysis. We will not accept a fluorescence in situ hybridization (FISH) test because it does not distinguish between the mosaic and non-mosaic forms of Down syndrome.

b. If a physician (see §§404.1513(a)(1) and 416.913(a)(1) of this chapter) has not signed the laboratory report of karyotype analysis, the evidence must also include a physician's statement that you have Down syndrome.

c. For purposes of 110.06A, we do not require evidence stating that you have the distinctive facial or other physical features of Down syndrome.

2. If we do not have a laboratory report of karyotype analysis documenting that you have non-mosaic Down syndrome, we may find you disabled under 110.06B or 110.06C.

a. Under 110.06B, we need a physician's report stating: (i) your karyotype diagnosis or evidence that documents your type of Down syndrome that is consistent with prior karyotype analysis (for example, reference to a diagnosis of "trisomy 21") and (ii)

that you have the distinctive facial or other physical features of Down syndrome. We do not require a detailed description of the facial or other physical features of the disorder. However, we will not find that your disorder meets 110.06B if we have evidence—such as evidence of functioning inconsistent with the diagnosis—that indicates that you do not have non-mosaic Down syndrome.

b. If we do not have evidence of prior karyotype analysis (you did not have testing, or you had testing but we do not have information from a physician about the test results), we will find that your disorder meets 110.06C if we have: (i) a physician's report stating that you have the distinctive facial or other physical features of Down syndrome and (ii) evidence that your functioning is consistent with a diagnosis of non-mosaic Down syndrome. This evidence may include medical or nonmedical information about your physical and mental abilities, including information about your development, education, work history, or the results of psychological testing. However, we will not find that your disorder meets 110.06C if we have evidence—such as evidence of functioning inconsistent with the diagnosis—that indicates that you do not have non-mosaic Down syndrome.

D. What are catastrophic congenital disorders? Some catastrophic congenital disorders, such as anencephaly, cyclopia, chromosome 13 trisomy (Patau syndrome or trisomy D), and chromosome 18 trisomy (Edwards' syndrome or trisomy E), are usually expected to result in early death. Others such as cri du chat syndrome (chromosome 5p

deletion syndrome) and the infantile onset form of Tay-Sachs disease interfere very seriously with development. We evaluate catastrophic congenital disorders under 110.08. The term "very seriously" in 110.08 has the same meaning as in the term "extreme" in §416.926a(e)(3) of this chapter.

E. What evidence do we need under 110.08?

We need one of the following to determine if your disorder meets 110.08A or B:

1. A laboratory report of the definitive test that documents your disorder (for example, genetic analysis or evidence of biochemical abnormalities) signed by a physician.
2. A laboratory report of the definitive test that documents your disorder that is not signed by a physician and a report from a physician stating that you have the disorder.
3. A report from a physician stating that you have the disorder with the typical clinical features of the disorder and that you had definitive testing that documented your disorder. In this case, we will find that your disorder meets 110.08A or B unless we have evidence that indicates that you do not have the disorder.
4. If we do not have the definitive laboratory evidence we need under E1, E2, or

E3, we will find that your disorder meets 110.08A or B if we have: (i) a report from a physician stating that you have the disorder and that you have the typical clinical features of the disorder, and (ii) other evidence that supports the diagnosis. This evidence may include medical or nonmedical information about your development and functioning.

5. For obvious catastrophic congenital anomalies that are expected to result in early death, such as anencephaly and cyclopia, we need evidence from a physician that demonstrates that the infant has the characteristic physical features of the disorder. In these rare cases, we do not need laboratory testing or any other evidence that confirms the disorder.

F. How do we evaluate mosaic Down syndrome and other congenital disorders that affect multiple body systems?

1. Mosaic Down syndrome. Approximately 2 percent of children with Down syndrome have the mosaic form. In mosaic Down syndrome, there are some cells with an extra copy of chromosome 21 and other cells with the normal two copies of chromosome 21. Mosaic Down syndrome can be so slight as to be undetected clinically, but it can also be profound and disabling, affecting various body systems.

2. Other congenital disorders that affect multiple body systems. Other congenital disorders, such as congenital anomalies, chromosomal disorders, dysmorphic syndromes,

inborn metabolic syndromes, and perinatal infectious diseases, can cause deviation from, or interruption of, the normal function of the body or can interfere with development. Examples of these disorders include both the juvenile and late-onset forms of Tay-Sachs disease, trisomy X syndrome (XXX syndrome), fragile X syndrome, phenylketonuria (PKU), caudal regression syndrome, and fetal alcohol syndrome. For these disorders and other disorders like them, the degree of deviation, interruption, or interference, as well as the resulting functional limitations and their progression, may vary widely from child to child and may affect different body systems.

3. Evaluating the effects of mosaic Down syndrome or another congenital disorder under the listings. When the effects of mosaic Down syndrome or another congenital disorder that affects multiple body systems are sufficiently severe we evaluate the disorder under the appropriate affected body system(s), such as musculoskeletal, special senses and speech, neurological, or mental disorders. Otherwise, we evaluate the specific functional limitations that result from the disorder under our other rules described in 110.00G.

G. What if your disorder does not meet a listing? 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. See §416.926 of this chapter. If your impairment(s) does not meet or medically equal a listing, we will consider whether it functionally equals the listings. See §§416.924a and 416.926a of this chapter. We use the

rules in §416.994a of this chapter when we decide whether you continue to be disabled.

110.01 Category of Impairments, Congenital Disorders That Affect Multiple
Body Systems

110.06 Non-mosaic Down syndrome (chromosome 21 trisomy or chromosome 21
translocation), documented by:

A. A laboratory report of karyotype analysis signed by a physician, or both a
laboratory report of karyotype analysis not signed by a physician and a statement by a
physician that the child has Down syndrome (see 110.00C1).

OR

B. A physician's report stating that the child has chromosome 21 trisomy or
chromosome 21 translocation consistent with karyotype analysis with the distinctive
facial or other physical features of Down syndrome (see 110.00C2a).

OR

C. A physician's report stating that the child has Down syndrome with the
distinctive facial or other physical features and evidence demonstrating that the child is

functioning at the level of a child with non-mosaic Down syndrome (see 110.00C2b).

110.08 A catastrophic congenital disorder (see 110.00D and 110.00E) with:

A. Death usually expected within the first months of life.

OR

B. Very serious interference with development or functioning.

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