Genetic and Congenital Disorders

Definition/ cut-off value

Hereditary or congenital condition at birth that causes physical or metabolic abnormality. The current condition must alter nutrition status metabolically, mechanically, or both. May include, but is not limited to, cleft lip or palate, Down's syndrome, thalassemia major, sickle cell anemia (not sickle cell trait), and muscular dystrophy.

Presence of genetic and congenital disorders diagnosed by a physician as self reported by applicant/participant/caregiver; or as reported or documented by a physician, or someone working under physician's orders.

Participant category and priority level

Category	Priority
Pregnant Women	I
Breastfeeding Women	I
Non-Breastfeeding Women	III, IV, V, or VI
Infants	I
Children	III

Justification

For women, infants, and children with these disorders, special attention to nutrition may be required to achieve adequate growth and development and/or to maintain health.

Severe cleft lip and palate anomalies commonly cause difficulty with chewing, sucking and swallowing, even after extensive repair efforts (5). Surgery is required for many gastrointestinal congenital anomalies. (Examples are trachea-esophageal fistula, esophageal atresia, gastroschisis, omphalocele, diaphragmatic hernia, intestinal atresia, and Hirschsprung's Disease.)

Impaired esophageal atresia and trachea-esophageal fistula can lead to feeding problems during infancy. The metabolic consequences of impaired absorption in short bowel-syndrome, depend on the extent and site of the resection or the loss of competence. Clinical manifestations of short bowel syndrome, include diarrhea, dehydration, edema, general malnutrition, anemia, dermatitis, bleeding tendencies, impaired taste, anorexia, and renal calculi. Total parenteral feedings are frequently necessary initially, followed by gradual and individualized transition to oral feedings. After intestinal

resection a period of adaptation by the residual intestine begins and may last as long as 12-18 months (3). Even after oral feedings are stabilized, close follow-up and frequent assessment of the nutritional status of infants with repaired congenital gastro-intestinal anomalies is recommended (5).

Sickle-cell anemia is an inherited disorder in which the person inherits a sickle gene from each parent. Persons with sickle-cell trait carry the sickle gene, but under normal circumstances are completely asymptomatic. Good nutritional status is important to individuals with sickle-cell anemia to help assume adequate growth (which can be compromised) and to help minimize complications of the disease since virtually every organ of the body can be affected by sickle-cell anemia (i.e., liver, kidneys, gall ladder, and immune system). Special attention should be given to assuring adequate caloric, iron, folate, vitamin E and vitamin C intakes as well as adequate hydration.

Muscular dystrophy is a familial disease characterized by progressive atrophy and wasting of muscles. Changes in functionality and mobility can occur rapidly and as a result children may gain weight quickly (up to 20 pounds in a 6 month period). Early nutrition education that focuses on foods to include in a balanced diet, limiting foods high in simple sugars and fat and increasing fiber intake can be effective in minimizing the deleterious effects of the disease.

References

1. American Dietetic Association, Pediatric Nutrition Practice Group. Pediatric manual of clinical dietetics. Chicago: Pediatric Nutrition Dietetic Practice Group, American Dietetic Association, 1998.

2. Ekvall S. Pediatric nutrition in chronic diseases and developmental disorders prevention, assessment, and treatment. New York: Oxford University Press 1993. p. 289-292.

3. Grand RJ, Sutphen JL, Dietz WH. Pediatric nutrition theory and practice. Boston: Butterworths, 1987.

4. Institute of Medicine. WIC nutrition risk criteria a scientific assessment. National Academy Press, Washington, D.C.; 1996.

5. Ohio Neonatal Nutritionists. Nutritional care for high risk newborns. Philapelphia, PA: G.F. Stickley Publishers, 1985.

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Clarification

Self-reporting of a diagnosis by a medical professional should not be confused with self-diagnosis, where a person simply claims to have or to have had a medical condition without any reference to professional diagnosis. A self-reported medical diagnosis ("My doctor says that I have/my son or daughter has…" Should prompt the CPA to validate the presence of the condition by asking more pointed questions related to that diagnosis.