# Congenital Disorders

## OBJECTIVE

The objective of this Clinical Practice Guideline (CPG) is to provide evidence-based practice recommendations for Rare Congenital Disorders. The CPG discusses medical and behavioral health implications. In addition, the CPG outlines the organizations that WellCare aligns with regarding Congenital Disorders and relevant Measureable Health Outcomes.

## OVERVIEW

Congenital disorders (or birth defects) are the result of structural changes in one or more parts of the body. They are present at birth and have serious, adverse effects on the health, development, and/or functional ability of the child.

**Structural birth defects** include:¹
- Cleft lip or cleft palate
- Heart defects, such as missing or misshaped valves
- Abnormal limbs, such as a clubfoot
- Neural tube defects (e.g., spina bifida and problems related to the development of the brain and spinal cord)

**Functional or developmental birth defects** include:¹
- **Nervous system or brain problems** – includes intellectual and developmental disabilities, behavioral disorders, speech or language difficulties, seizures, and movement trouble (e.g., Down syndrome, Prader-Willi syndrome, and Fragile X syndrome)
- **Sensory problems** – includes hearing loss and visual problems (e.g., blindness or deafness)
- **Metabolic disorders** – includes problems with certain chemical reactions in the body, such as conditions that limit the body’s ability to rid itself of waste materials or harmful chemicals (e.g., phenylketonuria and hypothyroidism)
- **Degenerative disorders** – includes conditions that might not be obvious at birth however the child’s health declines (e.g., muscular dystrophy and X-linked adrenoleukodystrophy [subject of the film "Lorenzo’s Oil."]) 

The most commonly diagnosed congenital disorders are noted below – for more information, visit the CDC’s page on Birth Defects [here].²

- Anencephaly
- Anophthalmia / Microphthalmia
- Anotia/Microtia
- Cleft Lip / Cleft Palate
- **Congenital Heart Defects**
  - Atrial Septal Defect
  - Atrioventricular Septal Defect
  - Coarctation of the Aorta
  - D-Transposition of the Great Arteries
  - Hypoplastic Left Heart Syndrome
  - Pulmonary Atresia
  - Tetralogy of Fallot
  - Total Anomalous Pulmonary Venous Return
  - Tricuspid Atresia
  - Truncus Arteriosus
  - Ventricular Septal Defect
- **Craniosynostosis**
- **Diaphragmatic Hernia**
- **Down Syndrome**
- **Encephalocele**
- **Esophageal Atresia**
- **Gastrochisis**
- **Hypospadias**
- **Microcephaly**
- **Omphalocele**
- **Spina Bifida**
- **Upper and Lower Limb Reduction Defects**

Birth defects affect one in every 33 babies (about 3% of all babies) born in the United States each year and are the leading cause of infant deaths, accounting for 20% of all infant deaths.³ The results of a 12-state study published by the *American Journal of Public Health* found the following:⁴
• Compared to non-Hispanic Whites, American Indians/Alaska Natives had a significantly higher occurrence of:
  o Anotia/Microtia (ear defects)
  o Cleft lip with or without cleft palate
  o Trisomy 18
  o Encephalocele (serious defect of the skull and brain)
  o Limb deficiency (when part or all of the arm or leg fails to form completely during pregnancy)
• Cubans and Asians (especially Chinese and Asian Indians) had significantly lower occurrence of many of the studied birth defects, compared to non-Hispanic whites.

Three main types of congenital disorders are explored in this CPG – cleft lip / palate, spina bifida, and congenital heart defects.

**Cleft Lip / Palate**

Children with a cleft lip or a cleft palate, depending on the size of the openings, may have problems eating and breathing. As they grow older, they may also have speech and language delays. Children with cleft lip or palate are also more likely to have ear infections, hearing loss, and problems with their teeth. Treatment for children with cleft lip or palate depends on the size of the cleft, the child's age and needs, and whether there are additional problems related to a genetic syndrome. A child with cleft lip or palate is often referred to a team of Specialists that may include an otolaryngologist, plastic surgeon, oral surgeon, speech pathologist, pediatric dentist, orthodontist, audiologist, pediatrician, nutritionist, and psychologist/social worker.5

**Spina Bifida**

Spina bifida (SB) is a neural tube defect (a disorder involving incomplete development of the brain, spinal cord, and/or their protective coverings) caused by the failure of the fetus's spine to close properly during the first month of pregnancy. Infants born with SB sometimes have an open lesion on their spine where significant damage to the nerves and spinal cord has occurred. The nerve damage is permanent, resulting in varying degrees of paralysis of the lower limbs. In addition to physical and mobility difficulties, most individuals have some form of learning disability. SB may also cause bowel and bladder complications, and many children with SB have hydrocephalus (excessive accumulation of cerebrospinal fluid in the brain). Treatment for the variety of effects of SB may include surgery, medication, and physical therapy. Many individuals with SB will need assistive devices such as braces, crutches, or wheelchairs. Ongoing therapy, medical care, and/or surgical treatments may be necessary to prevent and manage complications throughout the individual's life.

**Congenital Heart Defects**

There are many types of congenital heart defects ranging from simple to complex and critical. Simple defects (atrial septal defect and ventricular septal defects), may have no symptoms and may not require surgery. Complex or critical defects (hypoplastic left heart syndrome) may have severe, life-threatening symptoms. Babies born with a critical congenital heart defect typically have low levels of oxygen soon after birth and need surgery within the first year of life.

**Hierarchy of Support**

CPGs are updated annually or as necessary due to updates made to guidelines or recommendations by the American Academy of Pediatrics (AAP), American College of Obstetricians and Gynecologists (ACOG), and the Spina Bifida Foundation (SBF). When there are differing opinions noted by national organizations, WellCare will default to the member’s benefit structure as deemed by state contracts and Medicaid / Medicare regulations. If there is no specific language pertaining to Rare Congenital Disorders, WellCare will default (in order) to the following:

- National Committee for Quality Assurance (NCQA);
- United States Preventive Services Task Force (USPSTF), National Quality Strategy (NQS), Agency for Healthcare Research and Quality (AHRQ);
- Specialty associations, colleges, societies, etc. (e.g., American Academy of Family Physicians, American...
Links to websites within the CPGs are provided for the convenience of Providers. Listings do not imply endorsement by WellCare of the information contained on these websites. NOTE: All links are current and accessible at the time of MPC approval.

WellCare aligns with AAP, ACOG, and SBFO on the topic of Rare Congenital Disorders. Highlights from their respective publications are noted below.

**AMERICAN ACADEMY OF PEDIATRICS**

The American Academy of Pediatrics (AAP) and the American College of Cardiology (ACC) developed a joint statement which focuses on the care of and improvement of health outcomes for children with CHD. Providers such as the primary care provider (PCP) and the pediatric cardiologist would lead this effort. *The Care of Children With Congenital Heart Disease in Their Primary Medical Home* (available [here](#)) focuses on anticipated problems and places an emphasis on the role of PCP – particularly in the management of patients and their families through the life stages (prenatal to adolescence). Recommendations include:

- Promotion of care coordination and communication among family and care providers, especially during transitions.
- Advocating for caregiver support.
- Facilitating patient access to subspecialty care and medications.
- Staying current on pediatric basic and advanced life support and encouraging CPR training.
- Enhancing the neonatal history and physical exam with pulse oximetry screening.
- Encouraging growth and development by ensuring adequate nutrition.
- Awareness of infection prevention strategies, including special immunization needs and endocarditis prophylaxis regimens.
- Anticipating extra-cardiac organ involvement, complications of therapy and susceptibility to common childhood illnesses.
- Promotion of good nutrition and appropriate physical activity.
- Counseling against illicit drug, alcohol and tobacco use, unprotected sex and teen pregnancy.
- Facilitating transitioning to adult health care when appropriate.
- Supporting electronic health record (HER) accuracy and access to medical information for all providers.

**AMERICAN COLLEGE OF OBSTETRICIANS AND GYNECOLOGISTS (ACOG)**

The American College of Obstetricians and Gynecologists (ACOG) published two practice bulletins:

- *Prenatal Diagnostic Testing for Genetic Disorders* [7]
- *Screening for Fetal Aneuploidy* [8]

In addition, ACOG has published the following resources:

- *Prenatal Genetic Diagnostic Tests (FAQ 164)* [9]
- *Prenatal Genetic Screening Tests (FAQ 165)* [10]

**SPINA BIFIDA FOUNDATION**

The Spina Bifida Foundation published *Guidelines for the Care of People with Spina Bifida* are available [here](#). The guidelines are clear and evidence-based with information regarding treatment for those age birth through adulthood. In addition, 25 topics are included ranging from bowel management to sexuality to physical activity. [11]

**Evidence Based Practice**

**AGENCY FOR HEALTHCARE RESEARCH AND QUALITY (AHRQ)**

The Agency for Healthcare Research and Quality (AHRQ) has not published reports on this topic.

**MEASUREMENT OF COMPLIANCE**
WellCare is committed to adhering to the measures and standards published by the Centers for Medicare and Medicaid Services (CMS) and the National Committee for Quality Assurance (NCQA). Please reference WellCare’s Clinical Policy Guiding Document titled *Quality Improvement*.

NOTE: To access Clinical Policy Guiding Documents visit [www.wellcare.com](http://www.wellcare.com)—select the Provider tab, then “Tools” and “Clinical Guidelines”.

## Care Management

### Spina Bifida

The goals for Care Management are to support the Member’s / Member’s parent’s/guardian’s ability to self-promote their health, encourage healthy behaviors to minimize risks of disease and/or complications thereof, and remove barriers preventing the Member’s / Member’s family from achieving those goals. Primary symptoms for assessment and member education include:

- Fall/Injury precautions
- Assist with durable medical equipment and supplies
- Address barriers to access to care
- Bowel/bladder incontinence
- Providing education on decreasing risk of complications
- Counseling regarding psychosocial issues
- Provide education on resources for learning disabilities if applicable

### Congenital Heart Disease

The goals for Care Management are to support the Member’s / Member’s family’s ability to self-promote their health, encourage healthy behaviors to minimize risks of disease and/or complications thereof, and remove barriers preventing the Member’s / Member’s family from achieving those goals. Primary symptoms for assessment and member education include:

- Arrhythmia
- Blood clots
- Developmental disorders and delays. Children with congenital heart defects are more likely to have problems with behavior. They are also more likely to have speech and attention-deficit/hyperactivity disorders.
- Emotional health issues. Depression, anxiety, and post-traumatic stress disorder are common among people with congenital heart defects.
- Endocarditis, a type of heart inflammation
- Endocrine disorders, including thyroid problems, bone health issues, and diabetes. Problems with the hormones that deal with calcium can cause bone problems.
- Heart failure is the leading cause of death in adults with congenital heart defects. Some children with congenital heart defects develop heart failure.
- Kidney disease
- Liver disease
- Pneumonia is a leading cause of death in adults with congenital heart disease.
- Women with congenital heart defects have an increased risk of complications during pregnancy and childbirth.
- Pulmonary hypertension
- Stroke

### Cleft Palate

The goals for Care Management are to support the Member’s / Member’s family’s ability to self-promote their health, encourage healthy behaviors to minimize risks of disease and/or complications thereof, and remove barriers preventing the Member’s / Member’s family from achieving those goals. Primary symptoms for assessment and member education include:

- Assess and address concerns with breathing, coordinate care with specialist
- Assess and address nutritional status/problems with feeding
• Assist with coordinating care for speech
• Assist with locating providers and address barriers to care
• Assess for dental concerns and assist with locating providers
• Assess for and address emotional/behavioral health concerns

MEASURABLE HEALTH OUTCOMES

1. The member reports fewer or well-managed symptoms over a specific period after the start of Case Management engagement. Member-specific goals should reference member’s individual symptoms. Compare member’s symptom assessment responses, initial to subsequent assessments.
2. The member experiences no symptoms requiring acute medical care and intervention. Compare pre- and post-engagement utilization frequency. Monitor for ED and inpatient authorization/utilization related to the primary diagnosis. In absence of ED and inpatient utilization, authorizations and claims data, or to otherwise demonstrate less frequent need for acute medical intervention, CM may use Provider and/or Member narrative.

CASE MANAGEMENT GOALS

Case Goals should target specific care gaps and/or adherence issues, and measure the member’s progress towards self-management and adherence, which will lead to the targeted health outcomes above. Examples:
  • Member describes the use of fall/injury precautions over the last 30 days.
  • Member’s treatment demonstrates at least an 80% adherence rate of prescribed medications (verified by claims or member/provider narrative) over last 30 days.
  • Member describes signs and symptoms of infection and when to seek care from PCP/urgent care
  • Member obtains supplies needed to manage bowel/bladder incontinence
  • Specific for Members requiring hospitalization: Member participates in provider follow-up visit within 7 days of hospital discharge.

CASE MANAGEMENT OBJECTIVES

Spina Bifida

Case Management objectives should focus on improving the member’s self-management skills including:
  • Provide skills for coping, support systems, behavioral health referrals if needed for anxiety/depression
  • Coordinate care with PCP, Specialists, Therapies (Physical Therapy, Speech Therapy, Occupational Therapy)
  • Assist with obtaining durable medical equipment and supplies
  • Assess member/caregiver ability to complete activities of daily living (ADLs)
  • Assess safety/mobility
  • Provide education on signs and symptoms of urinary tract infection
  • Provide education and coordinate with provider for bowel and bladder incontinence
  • Provide education on avoiding complications of spina bifida
  • Assist with transition of care as member ages, assist with locating providers and appointments
  • Provide information on support groups/therapy/educational groups for family members of the member.

Congenital Heart Disease

Case Management objectives should focus on improving the member’s self-management skills including:
  • Coordinate care, including regular follow up with a pediatric cardiologist or an adult congenital heart specialist as directed
  • Following up with primary care doctor for routine exams
  • Taking medicines as prescribed to prevent complications
  • Going to the dentist for routine cleanings and oral care
  • Adults should go to medical centers that have specialized programs for adults with congenital heart disease.
  • Provide education and coaching on heart healthy diet, achieving healthy weight, exercise
  • Coordinate care for developmental delays (physical, occupational, speech therapy)
• Assess/address emotional/behavioral health concerns
• Provide education on birth control and pregnancy
• Assist with transition from pediatric to adult care
• Provide education on preventing complications:
  o **Anti-arrhythmics.** These drugs control arrhythmia and may be used for patients whose congenital heart defect causes arrhythmia.
  o **Antibiotics.** People with certain types of congenital heart defects may have an increased risk of infective endocarditis. Your doctor may recommend antibiotics to reduce the risk of infective endocarditis before dental procedures or other procedures that run the risk of introducing bacteria to the bloodstream. Good oral health also decreases the risk of infective endocarditis.
  o **Anticlotting medicines.** You may need to take anticoagulant, antiplatelet, and fibrinolytic medicines to treat blood clots or prevent blood clots from forming. These medicines are often prescribed long-term to people with artificial shunts and mechanical heart valves. Long-term use of warfarin, a common anticoagulant, may increase the risk of osteoporosis.
  o **Blood pressure medicines.** These drugs help control blood pressure. Common types of blood pressure medicines include diuretics, beta blockers, and angiotensin-converting enzyme (ACE) inhibitors.
  o **Pacemaker.** Pacemakers can be given to both children and adults with congenital heart defects to help control abnormal heart rhythms, also known as arrhythmias.
  o **Routine vaccinations** are especially important for children with congenital heart defects to help prevent abnormal heart rhythms. Adults with ongoing heart or immune problems should have a pneumococcal vaccination to prevent pneumonia and complications such as meningitis.
  o **Special care during surgery.** Be sure your doctor is aware of your congenital heart defect before any surgery, not just heart surgery. People with congenital heart defects are at higher risk of problems during surgery.
  o **Provide education regarding Training for sudden cardiac arrest.** Caregivers and family members can train in cardiopulmonary resuscitation (CPR) and using a type of defibrillator called an automated external defibrillator (AED).12
  o Provide education and assist with coordinating appointments and address barriers to care for required testing which may include: **Blood or urine tests** to monitor the function of organs affected by a congenital heart defect. **Spirometry** to measure how well the lungs are working. **Abdominal imaging** by ultrasound, magnetic resonance imaging (MRI), or computed tomography (CT) to look for liver disease.

### Cleft Palate

Case Management objectives should focus on improving the member’s self-management skills including:13

• Following up with primary care doctor for routine exams
• Coordinating care with Specialists
• Assess and address nutrition concerns
• Assess and address emotional concerns

### MEDICAL BEHAVIORAL INTEGRATION

Please note above Case Management Objectives regarding behavioral health.

### MEMBER EDUCATIONAL RESOURCES

Currently there are no Krames/StayWell Member educational materials utilized by WellCare Case Managers.

For additional information, please visit the CDC’s page on Birth Defects [here](#). Topics include facts, prevention, education on specific birth defects, statistics, and a variety of multimedia resources.2

For information on reducing the risk of birth defects from ACOG, click [here](#).14

For additional information on living with **Spina Bifida** please visit the following websites:

• [Centers for Disease Control and Prevention](#) 15
• March of Dimes  
• National Dissemination Center for Children with Disabilities via the United States Department of Education, Office of Special Education Programs  
• National Institute of Child Health and Human Information Resource Center  
• Spina Bifida Association  

For additional information on living with Congenital Heart Disease please visit the following websites:
• American Academy of Pediatrics  
• American Congenital Heart Association  
• American Heart Association  
• Centers for Disease Control and Prevention  
• Pediatric Congenital Heart Association  
• National Heart, Lung, and Blood Institute  
• March of Dimes  

For additional information on Cleft Palate please visit the following websites:
• American Cleft Palate-Craniofacial Association (ACPA)  
• Cleft Palate Foundation (CPF)  
• Cleft Lip & Palate Foundation of Smiles  

Related WellCare Guidelines

In addition to the information contained in this document, please reference the following CPG: Congenital Metabolic Disorders: HS-1060; Neonatal and Infant Health: HS-1072; and Special Health Care Needs for Children: HS-1061.

NOTE: Clinical Policies can be accessed by going to www.wellcare.com – select the Provider tab, then “Tools” and “Clinical Guidelines”.  

References  

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Medical Policy Committee Approval History

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