Genetic Testing for CHARGE Syndrome

Policy # 00393
Original Effective Date: 11/20/2013
Current Effective Date: 05/08/2023

Applies to all products administered or underwritten by Blue Cross and Blue Shield of Louisiana and its subsidiary, HMO Louisiana, Inc. (collectively referred to as the “Company”), unless otherwise provided in the applicable contract. Medical technology is constantly evolving, and we reserve the right to review and update Medical Policy periodically.

When Services Are Eligible for Coverage

Coverage for eligible medical treatments or procedures, drugs, devices or biological products may be provided only if:

- Benefits are available in the member’s contract/certificate, and
- Medical necessity criteria and guidelines are met.

Based on review of available data, the Company may consider genetic testing for CHARGE syndrome to confirm a diagnosis in an individual with signs/symptoms of CHARGE syndrome when a definitive diagnosis cannot be made with clinical criteria to be eligible for coverage** (see Policy Guidelines section).

When Services Are Considered Investigational

Coverage is not available for investigational medical treatments or procedures, drugs, devices or biological products.

Based on review of available data, the Company considers genetic testing for CHARGE syndrome in all other situations to be investigational.*

Policy Guidelines

A diagnosis of definitive CHARGE syndrome can be made clinically in individuals with all 4 major characteristics, or 3 major and 3 minor characteristics (Lalani et al [2012]). In individuals without the classical clinical criteria to diagnose CHARGE, in those with a milder phenotype, and/or in those with features that overlap with and cannot be distinguished from other syndromes, genetic testing may provide a definitive diagnosis.

Major characteristics include ocular coloboma, choanal atresia or stenosis, cranial nerve abnormality, and ear anomalies/deafness.
Minor characteristics include genital hypoplasia, hypogonadotropic hypogonadism, developmental delays, cardiac malformations, short stature, cleft lip and/or cleft palate, tracheoesophageal fistula, and distinctive CHARGE facial appearance, consisting of a prominent forehead and a prominent nasal bridge. Other, less frequent manifestations include kidney malformations, immunodeficiency, various limb abnormalities, scoliosis, dental problems, omphalocele, brain malformations, attention-deficit/hyperactivity disorder, and various behavioral problems.

This policy does not address preconception (carrier) testing and prenatal (in utero) testing.

Genetics Nomenclature Update
The Human Genome Variation Society nomenclature is used to report information on variants found in DNA and serves as an international standard in DNA diagnostics. It is being implemented for genetic testing medical evidence review updates starting in 2017 (see Table PG1). The Society’s nomenclature is recommended by the Human Variome Project, the Human Genome Organisation, and the Human Genome Variation Society itself.

The American College of Medical Genetics and Genomics (ACMG) and the Association for Molecular Pathology (AMP) standards and guidelines for interpretation of sequence variants represent expert opinion from both organizations, in addition to the College of American Pathologists. These recommendations primarily apply to genetic tests used in clinical laboratories, including genotyping, single genes, panels, exomes, and genomes. Table PG2 shows the recommended standard terminology - “pathogenic,” “likely pathogenic,” “uncertain significance,” “likely benign,” and “benign” - to describe variants identified that cause Mendelian disorders.

Table PG1. Nomenclature to Report on Variants Found in DNA

<table>
<thead>
<tr>
<th>Previous</th>
<th>Updated</th>
<th>Definition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mutation</td>
<td>Disease-associated variant</td>
<td>Disease-associated change in the DNA sequence</td>
</tr>
<tr>
<td>Variant</td>
<td>Change in the DNA sequence</td>
<td></td>
</tr>
</tbody>
</table>
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| Familial variant | Disease-associated variant identified in a proband for use in subsequent targeted genetic testing in first-degree relatives |

Table PG2. ACMG-AMP Standards and Guidelines for Variant Classification

<table>
<thead>
<tr>
<th>Variant Classification</th>
<th>Definition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pathogenic</td>
<td>Disease-causing change in the DNA sequence</td>
</tr>
<tr>
<td>Likely pathogenic</td>
<td>Likely disease-causing change in the DNA sequence</td>
</tr>
<tr>
<td>Variant of uncertain significance</td>
<td>Change in DNA sequence with uncertain effects on disease</td>
</tr>
<tr>
<td>Likely benign</td>
<td>Likely benign change in the DNA sequence</td>
</tr>
<tr>
<td>Benign</td>
<td>Benign change in the DNA sequence</td>
</tr>
</tbody>
</table>

ACMG: American College of Medical Genetics and Genomics; AMP: Association for Molecular Pathology.

**Genetic Counseling**

Genetic counseling is primarily aimed at individuals who are at risk for inherited disorders, and experts recommend formal genetic counseling in most cases when genetic testing for an inherited condition is considered. The interpretation of the results of genetic tests and the understanding of risk factors can be very difficult and complex. Therefore, genetic counseling will assist individuals in understanding the possible benefits and harms of genetic testing, including the possible impact of the information on the individual's family. Genetic counseling may alter the utilization of genetic testing substantially and may reduce inappropriate testing. Genetic counseling should be performed by an individual with experience and expertise in genetic medicine and genetic testing methods.

**Background/Overview**

**CHARGE Syndrome**

CHARGE syndrome is a rare genetic condition caused by variants of the CHD7 gene on chromosome 8q12.1. The letters of CHARGE syndrome correspond to clinical features: C = coloboma; H = heart defect; A = atresia choanae; R = retarded growth and development; G = genital hypoplasia; and E = ear anomalies/deafness. A number of other malformations are also common in
this condition. In particular, hypoplasia of the semicircular canals has emerged as a frequent and distinctive CHARGE malformation.

Newborns with CHARGE syndrome typically have several major congenital malformations that affect vision, hearing, cardiovascular function, growth, development, neurologic function, and overall well-being. Mortality is relatively high in neonates with bilateral choanal atresia, cyanotic cardiac malformations, central nervous system malformations, and/or tracheoesophageal fistula. In a 1998 series, the death rate was 20% in the first month of life and about 50% by 6 months of age. A formal 2005 epidemiologic study in Canada concluded that those who survived infancy were likely to have long-term survival. Morbidity is chronic and multisystemic. Cognitive outcomes are difficult to assess because both motor skills and language do not necessarily reflect intellect in this group. About 75% have some degree of intellectual disability. Among the 25% with normal intelligence, many are well-educated and live independently as adults. Morbidity can be reduced by early diagnosis and treatment.

Clinical Diagnosis
Investigators have debated the relative importance of certain clinical signs. Consequently, the diagnostic criteria for CHARGE syndrome have been repeatedly revised.

The complete phenotypic spectrum of CHARGE syndrome was only revealed after identification of the causative gene in 2004, and the phenotypic spectrum of the disease is highly variable.

A 2012 review proposed that the diagnosis of CHARGE syndrome be considered definite if an individual has 4 major characteristics, or 3 major and 3 minor characteristics, criteria initially proposed by Blake (the Blake criteria), and modified by Verloes. Individuals with 1 or 2 major characteristics and several minor characteristics would be considered to have probable or possible CHARGE syndrome (see Table 1).
# Table 1. Criteria for the Diagnosis of CHARGE Syndrome

<table>
<thead>
<tr>
<th>Characteristics</th>
<th>Prevalence</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Major</strong></td>
<td></td>
</tr>
<tr>
<td>Ocular coloboma, which may be manifest in the iris and/or the retina, choroid, and optic disc, and sometimes as microphthalmia.</td>
<td>80%-90%</td>
</tr>
<tr>
<td>Choanal atresia or stenosis, which may be unilateral or bilateral. Complete bilateral choanal atresia is a life-threatening emergency in a newborn because neonates are obligate nose breathers. Some CHARGE patients have a cleft palate, in which case the cleft fulfills this criterion.</td>
<td>50%-60%</td>
</tr>
<tr>
<td>CN abnormality, including hyposmia or anosmia (CN I), facial palsy (CN VII), auditory nerve hypoplasia causing sensorineural hearing loss (CN VIII), and/or swallowing problems with or without aspiration (CN IX and CN X).</td>
<td>70%-90%</td>
</tr>
<tr>
<td>Characteristic auditory manifestation of the external, middle, or inner ear. The external ear is often dysmorphic. A number of ossicular malformations of the middle ear are common. Sensorineural hearing loss is associated with a Mondini malformation of the cochlea, and vestibular dysfunction is caused by aplasia or hypoplasia of the semicircular canals in 95% of individuals with CHARGE. Temporal bone computed tomography is necessary to diagnose the cochlear and semicircular canal defects.</td>
<td>80%-100%</td>
</tr>
<tr>
<td><strong>Minor</strong></td>
<td></td>
</tr>
<tr>
<td>Genital hypoplasia in boys manifests as micropenis and cryptorchidism, and in girls as hypoplastic labia. Puberty may be delayed because of hypogonadotropic hypogonadism.</td>
<td>50%</td>
</tr>
<tr>
<td>Developmental delays, especially gross motor and language delays, which may be intrinsic qualities or caused by impaired balance, deafness, blindness, hypotonia, surgery, or other chronic illness.</td>
<td>100%</td>
</tr>
<tr>
<td>Congenital cardiac malformations.</td>
<td>80%</td>
</tr>
</tbody>
</table>
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<table>
<thead>
<tr>
<th>Characteristics</th>
<th>Prevalence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Short stature, often with postnatal onset.</td>
<td>75%</td>
</tr>
<tr>
<td>Cleft lip and/or cleft palate.</td>
<td>15%</td>
</tr>
<tr>
<td>Tracheoesophageal fistula.</td>
<td>15%</td>
</tr>
<tr>
<td>Distinctive CHARGE facial appearance, consisting of a prominent forehead and a prominent nasal bridge.</td>
<td>75%</td>
</tr>
</tbody>
</table>

CN: cranial nerve.

Other, less frequent manifestations include kidney malformations (25%), immunodeficiency, various limb abnormalities, scoliosis, dental problems, omphalocele, brain malformations, attention-deficit/hyperactivity disorder, and various behavioral problems. A systematic review and meta-analysis by Thomas et al (2022) investigated the prevalence of phenotypic characteristics and variability in CHARGE syndrome. Prevalence estimates were highest for developmental delay (84%), intellectual disability (64%), aggressive behavior (48%), sleep difficulties (45%), and self-injurious behavior (44%). Meta-regression indicated significant associations between intellectual disability and choanal atresia or inner ear anomalies, and sleep difficulties and growth deficiency or gross motor difficulties.

The diagnosis of CHARGE syndrome is primarily clinical, based on use of the diagnostic criteria above.

External ear anomalies, abnormalities of cranial nerve function, semicircular canal hypoplasia, and gross motor delays seem to be consistent phenotypic manifestations in CHARGE syndrome, but one-third of patients with CHARGE syndrome will lack choanal atresia and/or ocular coloboma, with the most mildly affected showing only abnormal ears and a balance disturbance. Consequently, CHARGE syndrome can closely resemble several other genetic and teratogenic conditions, such as the 22q11.2 deletion syndrome, Kallmann syndrome, VACTERL association, Kabuki syndrome, renal coloboma syndrome, cat-eye syndrome, Joubert syndrome, branchio-oto-renal syndrome, and retinoic embryopathy. In 1 patient with velo-cardio-facial syndrome in whom the chromosome 22q11.2 microdeletion was ruled out, a CHD7 variant was documented. Several patients with Kallmann syndrome were found to have CHD7 disease-associated variants.
In recognition of this expanding CHARGE phenotype, Bergman et al (2011) proposed a revision of cardinal and supporting features and suggested that CHD7 testing be offered to individuals on the milder end of the phenotypic spectrum. Their algorithmic approach to diagnosis also incorporated temporal bone computed tomography scans as an important but not necessary component of the diagnostic workup. Although CHARGE syndrome is most often related to a sporadic disease-associated variant, some investigators (2014) have proposed that family history (any first-degree relative with at least 1 major feature of CHARGE) be incorporated into the clinical diagnosis of CHARGE syndrome as a major diagnostic criterion.

Genetic Etiology
In 2014, certain variants of the CHD7 gene, which encodes chromodomain helicase DNA binding protein, were found to cause CHARGE syndrome. In mouse models, the CHD7 gene has been associated with neural crest migration. Almost all pathogenic variants have proven to be single nucleotide variants, though on rare occasions, there may be a chromosomal translocation with a breakpoint within the CHD7 gene. Microdeletions, as would be detected with chromosome microarray analysis, are rare and probably occur in no more than 2% of individuals.

CHARGE syndrome does not have sex-linked expression and hence affects males and females equally. Most instances of CHARGE syndrome are sporadic events in a family and appear to be caused by de novo CHD7 disease-associated variants. On rare occasions, CHARGE can be inherited as an autosomal dominant condition. Individuals with CHARGE who reproduce have a 50% chance of transmitting the variant to their offspring. Recurrence in siblings because of germline mosaicism has also been reported. The prevalence of CHARGE syndrome is estimated at 1 in 8500 live births.

Genetic testing for variants of CHD7 is available from several commercial laboratories and is generally performed through Sanger sequence analysis. If no disease-associated variant is identified by Sanger sequencing, deletion and duplication analysis can be performed to identify large deletions.

Treatment
Management (medical or surgical) is based on presentation timing, features, and severity. Extensive management guidelines have been developed for CHARGE syndrome. They include periodic examinations and treatment by ophthalmology, otolaryngology, audiology, occupational therapy, speech therapy, gastroenterology, endocrinology, cardiology, neurology, developmental pediatrics, and genetics. Routine investigations would include choanal computed tomography, nasal endoscopy,
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Brainstem auditory-evoked responses, temporal bone computed tomography, swallowing studies, renal ultrasound, gonadotropin testing, echocardiography, brain magnetic resonance imaging, growth hormone testing, and genetic counseling. Immunologic assessment should be considered, particularly if patients have recurrent lung or ear infections. Based on evaluation of immune dysfunction in children with CHARGE syndrome, Wong et al (2015) recommended immunologic evaluation of patients with CHARGE syndrome who have recurrent infections. Many of these resources might be provided in due course for a child with multiple congenital anomalies in the absence of an exact etiologic diagnosis. However, a number of specific investigations and therapies might not be considered unless CHARGE syndrome has been definitively diagnosed on a clinical basis or, for mildly affected individuals, as the result of genetic testing.

FDA or Other Governmental Regulatory Approval

U.S. Food and Drug Administration (FDA)
Clinical laboratories may develop and validate tests in-house and market them as a laboratory service; laboratory-developed tests must meet the general regulatory standards of the Clinical Laboratory Improvement Amendments (CLIA). Genetic tests for CHARGE syndrome are available under the auspices of the CLIA. Laboratories that offer laboratory-developed tests must be licensed by the CLIA for high-complexity testing. To date, the U.S. Food and Drug Administration has chosen not to require any regulatory review of this test.

Rationale/Source

This medical policy was developed through consideration of peer-reviewed medical literature generally recognized by the relevant medical community, U.S. Food and Drug Administration approval status, nationally accepted standards of medical practice and accepted standards of medical practice in this community, technology evaluation centers, reference to federal regulations, other plan medical policies, and accredited national guidelines.

CHARGE syndrome is a rare genetic condition associated with multiple congenital anomalies. In many individuals, the diagnosis can be made based on clinical findings. However, the phenotype of the disease is highly variable, and some patients do not fulfill the criteria for a definitive diagnosis by clinical findings. Sequence analysis of the CHD7 gene detects variants in most individuals with CHARGE syndrome.
Summary of Evidence

For individuals who have signs and/or symptoms of CHARGE syndrome who receive genetic testing for variants in the CHD7 gene, the evidence includes case series. Relevant outcomes are overall survival, test accuracy and validity, symptoms, morbid events, functional outcomes, quality of life, and resource utilization. Although the clinical sensitivity of CHD7 variant testing cannot be specifically defined, over 90% of patients who fulfill the Blake or Verloes criteria for CHARGE syndrome have a CHD7 variant. A definitive diagnosis may end the need for additional testing in the etiologic workup and direct patient care according to established clinical management guidelines for CHARGE syndrome, including referrals to appropriate specialists, treatment of manifestations, prevention of secondary complications, and surveillance. The evidence is sufficient to determine that the technology results in an improvement in the net health outcome.

Supplemental Information

Practice Guidelines and Position Statements
Guidelines or position statements will be considered for inclusion in ‘Supplemental Information’ if they were issued by, or jointly by, a US professional society, an international society with US representation, or National Institute for Health and Care Excellence (NICE). Priority will be given to guidelines that are informed by a systematic review, include strength of evidence ratings, and include a description of management of conflict of interest.

Bergman et al (2011) proposed guidelines for CHD7 analysis and stated that, while the diagnosis of CHARGE syndrome remains primarily a clinical diagnosis (Table 1), molecular testing can confirm the diagnosis in mildly affected patients.

U.S. Preventive Services Task Force Recommendations
Not applicable.

Medicare National Coverage
There is no national coverage determination. In the absence of a national coverage determination, coverage decisions are left to the discretion of local Medicare carriers.
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Ongoing and Unpublished Clinical Trials
A search of ClinicalTrials.gov in December 2022 did not identify any ongoing or unpublished trials that would likely influence this review.

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Policy History

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11/07/2013 Medical Policy Committee review
11/06/2014 Medical Policy Committee review
08/03/2015 Coding update: ICD10 Diagnosis code section added; ICD9 Procedure code section removed.
10/29/2015 Medical Policy Committee review
11/16/2015 Medical Policy Implementation Committee approval. Coverage eligibility unchanged

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11/03/2016 Medical Policy Committee review
11/16/2016 Medical Policy Implementation Committee approval. Coverage eligibility unchanged
01/01/2017 Coding update: Removing ICD-9 Diagnosis Codes
11/02/2017 Medical Policy Committee review
11/15/2017 Medical Policy Implementation Committee approval. Coverage eligibility unchanged
11/08/2018 Medical Policy Committee review
11/07/2019 Medical Policy Committee review
12/10/2019 Coding update
04/02/2020 Medical Policy Committee review
04/08/2020 Medical Policy Implementation Committee approval. Coverage eligibility unchanged.
04/01/2021 Medical Policy Committee review
04/14/2021 Medical Policy Implementation Committee approval. Coverage eligibility unchanged.
04/07/2022 Medical Policy Committee review
04/13/2022 Medical Policy Implementation Committee approval. Coverage eligibility unchanged.
04/06/2023 Medical Policy Committee review
04/12/2023 Medical Policy Implementation Committee approval. Replaced “a patient” with “an individual” in the coverage section. Coverage eligibility unchanged.
06/07/2023 Coding update
08/08/2023 Coding update

Next Scheduled Review Date: 04/2024

Coding
The five character codes included in the Blue Cross Blue Shield of Louisiana Medical Policy Coverage Guidelines are obtained from Current Procedural Terminology (CPT®)®, copyright 2022

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by the American Medical Association (AMA). CPT is developed by the AMA as a listing of descriptive terms and five character identifying codes and modifiers for reporting medical services and procedures performed by physician.

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Codes used to identify services associated with this policy may include (but may not be limited to) the following:

<table>
<thead>
<tr>
<th>Code Type</th>
<th>Code</th>
</tr>
</thead>
<tbody>
<tr>
<td>CPT</td>
<td>81407</td>
</tr>
<tr>
<td></td>
<td>Delete code effective 08/01/2023: 0401U</td>
</tr>
<tr>
<td>HCPCS</td>
<td>No codes</td>
</tr>
<tr>
<td>ICD-10 Diagnosis</td>
<td>All related diagnoses</td>
</tr>
</tbody>
</table>

*Investigational – A medical treatment, procedure, drug, device, or biological product is Investigational if the effectiveness has not been clearly tested and it has not been incorporated into standard medical practice. Any determination we make that a medical treatment, procedure, drug, device, or biological product is Investigational will be based on a consideration of the following:

A. Whether the medical treatment, procedure, drug, device, or biological product can be lawfully marketed without approval of the U.S. Food and Drug Administration (FDA) and

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whether such approval has been granted at the time the medical treatment, procedure, drug, device, or biological product is sought to be furnished; or

B. Whether the medical treatment, procedure, drug, device, or biological product requires further studies or clinical trials to determine its maximum tolerated dose, toxicity, safety, effectiveness, or effectiveness as compared with the standard means of treatment or diagnosis, must improve health outcomes, according to the consensus of opinion among experts as shown by reliable evidence, including:
   1. Consultation with technology evaluation center(s);
   2. Credible scientific evidence published in peer-reviewed medical literature generally recognized by the relevant medical community; or
   3. Reference to federal regulations.

**Medically Necessary (or “Medical Necessity”)** - Health care services, treatment, procedures, equipment, drugs, devices, items or supplies that a Provider, exercising prudent clinical judgment, would provide to a patient for the purpose of preventing, evaluating, diagnosing or treating an illness, injury, disease or its symptoms, and that are:
   A. In accordance with nationally accepted standards of medical practice;
   B. Clinically appropriate, in terms of type, frequency, extent, level of care, site and duration, and considered effective for the patient's illness, injury or disease; and
   C. Not primarily for the personal comfort or convenience of the patient, physician or other health care provider, and not more costly than an alternative service or sequence of services at least as likely to produce equivalent therapeutic or diagnostic results as to the diagnosis or treatment of that patient's illness, injury or disease.

For these purposes, “nationally accepted standards of medical practice” means standards that are based on credible scientific evidence published in peer-reviewed medical literature generally recognized by the relevant medical community, Physician Specialty Society recommendations and the views of Physicians practicing in relevant clinical areas and any other relevant factors.

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NOTICE: If the Patient’s health insurance contract contains language that differs from the BCBSLA Medical Policy definition noted above, the definition in the health insurance contract will be relied upon for specific coverage determinations.
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