

<b>Department of Origin:</b> Integrated Healthcare Services	<b>Effective Date:</b> 03/05/24
<b>Approved by:</b> Medical Policy Quality Management Subcommittee	<b>Date Approved:</b> 03/05/24
<b>Clinical Policy Document:</b> Genetic Testing, Comparative Genomic Hybridization/ Chromosomal Microarray - Non-Oncology	<b>Replaces Effective Clinical Policy Dated:</b> 06/06/23
<b>Reference #:</b> MC/L015	<b>Page:</b> 1 of 10

#### **PURPOSE:**

The intent of this clinical policy is to ensure care is medically necessary.

Please refer to the member's benefit document for specific information. To the extent there is any inconsistency between this policy and the terms of the member's benefit plan or certificate of coverage, the terms of the member's benefit plan document will govern.

#### **POLICY:**

Benefits must be available for health care services. Health care services must be ordered by a provider. Licensed Genetic Counselors may also order genetic tests if it is within the scope of practice of their state licensure. Health care services must be medically necessary, applicable conservative treatments must have been tried, and the most cost-effective alternative must be requested for coverage consideration.

#### **GUIDELINES:**

Medical Necessity Criteria – Must satisfy any of the following: I - IV

- I. Comparative genomic hybridization (CGH) testing for chromosomal abnormalities in individuals is considered medically necessary when the request meets all of the following: A - D
  - A. *A health care professional trained in genetics*, independent of the laboratory performing the testing, has reviewed and documented family history, advised the parent/legal guardian of the potential harms of the testing and implications of the test results, and obtained written informed consent; and
  - B. If warranted, biochemical tests for metabolic disease have been performed and results are non-diagnostic; and
  - C. CGH testing is requested for one of the following: 1 - 4
    1. Nonsyndromic global developmental delay or intellectual disability (DD/ID); or
    2. Autism spectrum disorder (ASD); or
    3. Multiple congenital abnormalities (MCA) not specific to a well-defined genetic syndrome. (See Attachment A); or
    4. Isolated severe congenital heart disease.
  - D. The results of the genetic testing have the potential to impact the clinical management of the member.
- II. Comparative genomic hybridization testing in the prenatal setting is considered medically necessary for the following: A, and one of B - D
  - A. *A health care professional trained in genetics*, independent of the laboratory performing the testing, has reviewed and documented family history, advised the parent/legal guardian of the potential harms of the testing and implications of the test results, and obtained written informed consent; and

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- B. The member is pregnant with a fetus with one or more major structural abnormalities identified on ultrasound, fetal magnetic resonance imaging; or
  - C. The member is undergoing invasive prenatal testing (ie, amniocentesis, chorionic villus sampling or fetal tissue sampling); or
  - D. Evaluation of fetal death (stillbirth) at one of the following: 1 - 2
    - 1. 20 weeks or greater of gestation; or
    - 2. A weight greater than or equal to 350 grams if the gestational age is not known.
- III. Comparative genomic hybridization testing when member is a prospective parent or carrier testing (equal to or greater than age 12) – must have all of the following: A - D
- A. A *health care professional trained in genetics*, independent of the laboratory performing the testing, has reviewed and documented family history, advised the parent/legal guardian of the potential harms of the testing and implications of the test results, and obtained written informed consent; and
  - B. CGH testing of a previous fetus or child confirms a genetic condition or syndrome that puts future children at high risk for the specific inheritable disease, sickness, or defect; and
  - C. Conventional cytogenetic genetic testing is not adequate; and
  - D. Outcome of testing is required to determine carrier status of inherited disorders and to guide subsequent reproductive decisions.
- IV. Comparative genomic hybridization testing for chromosomal abnormalities in *neonates* is medically necessary as a first-line test when the *neonate* has multiple anomalies not specific to a well-defined genetic syndrome.

#### **EXCLUSIONS (not limited to):**

Refer to member's Certificate of Coverage or Summary Plan Description

CGH for all other indications is considered investigative (see Investigative List)

#### **DEFINITIONS:**

##### Copy Number Variants (CNVs):

An alteration of the DNA of a genome that results in the cell having an abnormal number of copies of one or more sections of the DNA.

##### Cytogenetics:

A branch of genetic science that focuses on the study of the structure and function of the cell, especially the chromosomes. Cytogenetics includes but is not limited to G-banded karyotyping, fluorescent in situ hybridization (FISH) and comparative genomic hybridization (CGH). Conventional cytogenetic testing is used to identify balanced rearrangements (eg, translocations or inversions), alterations in chromosome structure, sequence alterations, copy number changes (deletion, duplication and amplification), single-

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base pair mutation, 20% or lower level of mosaicism, and some types of polyploidy, including triploidy and tetraploidy. Conventional cytogenetic tests identify known genetic abnormalities associated with specific clinical syndromes. These tests may be used when a specific clinical syndrome is suspected.

#### FISH:

An established technique that labels specific regions of deoxyribonucleic acid (DNA), using sequence specific oligonucleotides (ie, short sequences of DNA) to identify chromosomal deletions, additions or rearrangements. Because FISH uses individual probes, it reveals DNA aberrations of only the probe-targeted segments. Locus-specific FISH detects subtelomeric and interstitial submicroscopic chromosomal arrangements (usually 3–5 megabases [Mb] in size) associated with particular phenotypes.

#### G-banded Karyotyping:

A molecular chromosome analysis technique which employs Giemsa dye to stain DNA strands. This method is indicated for evaluation of specific chromosome disorders, such as Down syndrome, sex chromosome abnormalities, and trisomy 13/18.

#### Health care professionals trained in genetics:

A genetics professional has experience and an educational background in genetics, counseling, and hereditary syndromes to provide accurate risk assessment and empathetic genetic counseling to patients and their families. Genetics professionals include people certified in any of the following ways:

- American Board of Genetic Counseling (ABGC) or American Board of Medical Genetics and Genomics (ABMGG) board certified/board eligible<sup>21</sup> or a licensed genetic counselor
- Advanced Genetics Nursing Certification (AGN-BC)<sup>21</sup>
- Advanced Clinical Genomics Nurse (ACGN) credential<sup>21</sup>
- Clinical Genomics Nurse (CGN) certification<sup>21</sup>
- Cancer Genetic Risk Assessment (CGRA) certification<sup>21</sup>
- Advanced practice oncology nurse or physician assistant with specialized education in cancer genetics and hereditary cancer predisposition syndromes<sup>21</sup>
- Board-certified/board-eligible physician with experience in cancer genetics (defined as education resulting in a certification and undergoing ongoing continuing medical education in cancer genetics and hereditary cancer predisposition syndromes)<sup>21</sup>
- A registered nurse with specialized education in cancer genetics and hereditary cancer predisposition syndromes (defined as education resulting in a certification and undergoing ongoing continuing medical education in cancer genetics and hereditary cancer predisposition syndromes)<sup>21</sup>
- Board-certified specialty care physician with experience in the diagnosis and treatment of the hereditary condition, eg, cardiologist ordering genetic testing for hypertrophic cardiomyopathy

#### Karyotypes:

The number and appearance of chromosomes under a light microscope.

#### Neonate:

A newborn infant equal to or less than four weeks of age

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## BACKGROUND:

Comparative genomic hybridization (CGH) or chromosomal microarray analysis is a laboratory method to aid in the detection of chromosomal imbalances. It allows for the detection of alterations (copy number variants or CNVs) in the genomic content of an individual. The technique works by comparing the DNA content of the individual with a normal control individual to identify pathogenic CNVs that may be responsible for the suspected disorder. Tens of thousands to millions of different DNA fragments (probes) are attached to identifiable locations on a glass slide or gene chip. Array CGH (aCGH) is a variation of CGH that detects chromosomal abnormalities at a higher resolution than conventional CGH, or chromosome-based CGH.

The three basic types of CGH are bacterial artificial chromosomes (BAC), oligonucleotide (oligo) and single nucleotide polymorphism (SNP) arrays. Arrays are often described as targeted or whole genome or genome-wide. Targeted arrays are high-resolution and contain only specific sections or regions of DNA containing known, clinically significant CNVs. Whole genome or genome-wide arrays cover the entire genome at varying levels of resolution. Whole genome arrays detect known CNVs, like targeted arrays, and may also detect the discovery of new CNVs.

IHC involves designing monoclonal antibodies that bind to the molecule being assessed. Formalin-fixed paraffin-embedded tissue is stained with the antibodies and the expression of the protein is assessed under a microscope. FISH is an established technique that labels specific regions of deoxyribonucleic acid (DNA), using sequence specific oligonucleotides (i.e., short sequences of DNA) to identify chromosomal deletions, additions or rearrangements. Because FISH uses individual probes, it reveals DNA aberrations of only the probe-targeted segments. Locus-specific FISH detects subtelomeric and interstitial submicroscopic chromosomal arrangements (usually 3–5 megabases [Mb] in size) associated with particular phenotypes. When high resolution G-banding is used, chromosomes are first treated with trypsin, an enzyme that degrades proteins. The chromosomes are then stained with Giemsa which produces a banding pattern of light and dark stripes enabling identification of each chromosome. G-band karyotyping is limited to a resolution of 5–10 Mb. PCR is an established laboratory method used to make numerous copies of a specific DNA sequence, utilizing pairs of oligonucleotide primers to replicate and alternate rounds of DNA. Real-time polymerase chain reaction, also called quantitative real time polymerase chain reaction (Q-PCR/qPCR/qrt-PCR) or kinetic polymerase chain reaction (KPCR), is a PCR technology used to simultaneously amplify and quantify the targeted DNA molecule. In reverse transcriptase PCR (RT-PCR) an RNA strand is reverse transcribed into its DNA complement (cDNA). Methylation-specific PCR (MSP) assesses the methylation status of DNA (American Association of Clinical Chemistry [AACC], 2010; Kibel and Reiter, 2007).

Conventional cytogenetic testing is used to identify balanced rearrangements (e.g., translocations or inversions), alterations in chromosome structure, sequence alterations, copy number changes (deletion, duplication and amplification), single-base pair mutation, 20% or lower level of mosaicism, and some types of polyploidy, including triploidy and tetraploidy. Conventional cytogenetic tests identify known genetic abnormalities associated with specific clinical syndromes. These tests may be used when a specific clinical syndrome is suspected.

DSM-5 Diagnostic Criteria for Autism Spectrum Disorders is available at the following website:  
<http://www.autismspeaks.org/what-autism/diagnosis/dsm-5-diagnostic-criteria>

DSM-5 Diagnostic Criteria for Intellectual Disability is available at the following website:  
<http://aaidd.org/intellectual-disability/definition>

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DSM-5 defines global developmental delay (GDD) as occurring in children less than five years of age who fail to meet expected developmental milestones in multiple areas of functioning. Developmental milestones are available at the following website:

<http://www.cdc.gov/ncbddd/actearly/milestones/index.html>

Major defects are structural abnormalities that affect the way a person looks and require medical and/or surgical treatment. Minor defects are abnormalities that do not cause serious health or social problems. When multiple birth defects occur together and have a similar cause, they are called syndromes. If two or more defects tend to appear together but do not share the same cause, they are called associations.

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Prior Authorization: Yes, per network provider agreement, outpatient setting.

#### **CODING:**

CPT®

81228 Cytogenomic (genome-wide) analysis for constitutional chromosomal abnormalities; interrogation of genomic regions for copy number variants, comparative genomic hybridization [CGH] microarray analysis

81229 Cytogenomic (genome-wide) analysis for constitutional chromosomal abnormalities; interrogation of genomic regions for copy number and single nucleotide polymorphism (SNP) variants, comparative genomic hybridization (CGH) microarray analysis

81349 Cytogenomic (genome-wide) microarray analysis for constitutional chromosomal abnormalities; interrogation of genomic regions for copy number and loss of heterozygosity variants, low-pass sequencing analysis

0209U Cytogenomic constitutional (genome-wide) analysis; interrogation of genomic regions for copy number changes and areas of homozygosity for chromosomal abnormalities

S3870 Comparative genomic hybridization (CGH) microarray testing for developmental delay, autism spectrum disorders and/or intellectual disability

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### Attachment A

Major Congenital Malformations		
<b>Head and Craniofacial Structures</b>	<u>Skull</u> <ul style="list-style-type: none"> <li>Anencephaly</li> <li>Encephalocele (occipital, frontal)</li> <li>Holoprosencephaly</li> <li>Hydrocephaly</li> </ul> <u>Eyes</u> <ul style="list-style-type: none"> <li>Microphthalmia</li> <li>Anophthalmia</li> <li>Colobomas (iris, retina)</li> </ul>	<u>Ears</u> <ul style="list-style-type: none"> <li>Microtia (types II through IV)</li> </ul> <u>Mouth and throat</u> <ul style="list-style-type: none"> <li>Cleft lip</li> <li>Cleft palate</li> <li>Severe micrognathia (Robin sequence)</li> <li>Macro- or microglossia</li> </ul>
<b>Neck</b>	<ul style="list-style-type: none"> <li>Cystic hygroma</li> </ul>	
<b>Chest</b>	<ul style="list-style-type: none"> <li>Pectus excavatum</li> <li>Absent or hypoplastic clavicles</li> </ul>	
<b>Back</b>	<ul style="list-style-type: none"> <li>Meningomyelocele</li> <li>Spina bifida</li> </ul>	
<b>Abdomen</b>	<ul style="list-style-type: none"> <li>Omphalocele</li> <li>Gastroschisis</li> </ul>	
<b>Genitalia</b>	<ul style="list-style-type: none"> <li>Ambiguous genitalia</li> </ul>	
<b>Extremities</b>	<u>Arms</u> <ul style="list-style-type: none"> <li>Absent or limb deficiencies</li> </ul>	<u>Hands and Feet</u> <ul style="list-style-type: none"> <li>Polydactyly, complete syndactyly, polysyndactyly</li> <li>Absent digits</li> <li>Ectrodactyly</li> </ul>
<b>Cardiovascular and great vessels</b>	<ul style="list-style-type: none"> <li>Tetralogy of Fallot</li> <li>Truncus arteriosus</li> <li>Hypoplastic left heart</li> <li>Ventricular or atrial septal defect</li> </ul>	<ul style="list-style-type: none"> <li>Transposition of the great vessels</li> <li>Interrupted aortic arch type B</li> <li>Total anomaly of pulmonary venous return</li> <li>Hypoplasia or coarctation of the aorta</li> </ul>

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#### Attachment A (continued)

Minor Congenital Malformations		
<b>Head and craniofacial Structures</b>	<u>Skull</u> <ul style="list-style-type: none"> <li>Abnormal hair whorls (absence, more than two)</li> <li>Frontal bossing</li> <li>Plagiocephaly</li> <li>Flat occiput</li> <li>Metopic fontanel</li> </ul> <u>Eyes</u> <ul style="list-style-type: none"> <li>Epicanthal folds</li> <li>Hypotelorism</li> <li>Hypertelorism</li> <li>Upslanting or downslanting palpebral fissures</li> <li>Short palpebral fissures</li> <li>Synophrys</li> <li>Ptosis</li> </ul>	<u>Ears</u> <ul style="list-style-type: none"> <li>Ear lobe - attached, creases, notches, or bifid</li> <li>Small ears (type I microtia)</li> <li>Lop ear</li> <li>Cup-shaped ear</li> <li>Protruding ear</li> <li>Ear tags</li> <li>Preauricular sinuses</li> </ul> <u>Nose</u> <ul style="list-style-type: none"> <li>Flat bridge</li> <li>Anteverted nostrils</li> <li>Philtrum long, short, flat</li> </ul> <u>Mouth and jaw</u> <ul style="list-style-type: none"> <li>Microstomia</li> <li>Macrostomia</li> <li>Bifid uvula</li> <li>Multiple frenula</li> <li>Micrognathia</li> <li>Retrognathia</li> </ul>
<b>Neck</b>	<ul style="list-style-type: none"> <li>Short neck</li> <li>Webbing</li> </ul>	<ul style="list-style-type: none"> <li>Redundant skin</li> <li>Branchial sinuses</li> </ul>
<b>Chest</b>	<ul style="list-style-type: none"> <li>Extra nipples</li> <li>Widely spaced nipples</li> <li>Low-placed nipples</li> </ul>	
<b>Back</b>	<ul style="list-style-type: none"> <li>Sacral dimple</li> </ul>	
<b>Genitalia</b>	<ul style="list-style-type: none"> <li>Shawl scrotum</li> <li>Vaginal tags</li> <li>Minor hypospadias</li> </ul>	
<b>Extremities</b>	<u>Arms</u> <ul style="list-style-type: none"> <li>Cubitus valgus</li> <li>Dimples over major joints</li> </ul> <u>Feet</u> <ul style="list-style-type: none"> <li>Partial syndactyly between two to three toes</li> <li>Nail hypoplasia</li> <li>Prominence of the heels</li> <li>Overlapping digits</li> </ul>	<u>Hands</u> <ul style="list-style-type: none"> <li>Fifth finger clinodactyly</li> <li>Single transverse palmar crease</li> <li>Bridge crease</li> <li>Tapered fingers</li> <li>Nail hypoplasia</li> <li>Persistent finger pads (fetal pads)</li> </ul>
<b>Skin</b>	<ul style="list-style-type: none"> <li>Nevi</li> <li>Hypo- or hyperpigmented macules</li> <li>Hemangioma</li> </ul>	

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## Attachment B

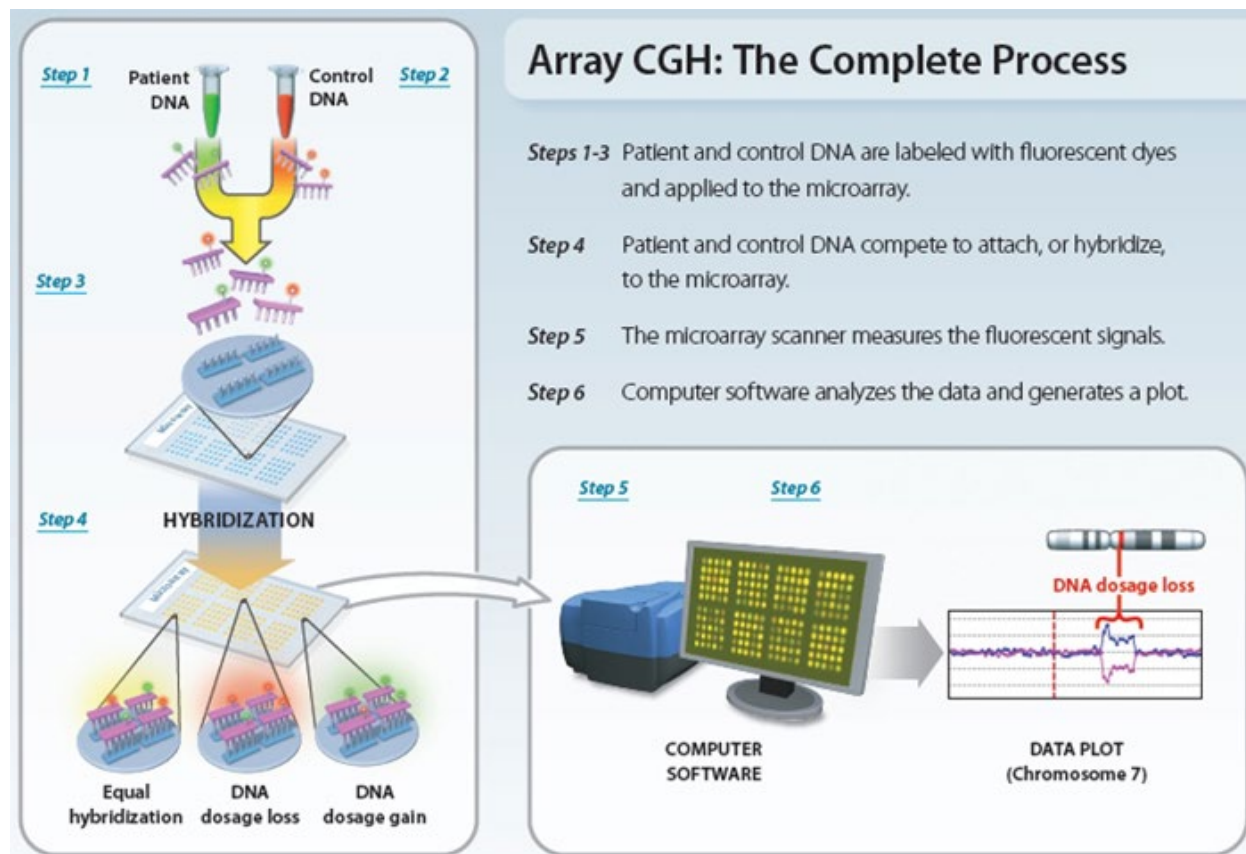


Diagram of the microarray-based comparative genomic hybridization (aCGH) process Source: [nature.com](https://www.nature.com)

# Nondiscrimination & Language Access Policy



Discrimination is Against the Law. Aspirus Health Plan, Inc. complies with applicable Federal civil rights laws and does not discriminate on the basis of race, color, national origin, age, disability, or sex, (including sex characteristics, including intersex traits; pregnancy or related conditions; sexual orientation, gender identity and sex stereotypes), consistent with the scope of sex discrimination described at 45 CFR § 92.101(a)(2). Aspirus Health Plan, Inc. does not exclude people or treat them less favorably because of race, color, national origin, age, disability, or sex.

Aspirus Health Plan, Inc.:

Provides people with disabilities reasonable modifications and free appropriate auxiliary aids and services to communicate effectively with us, such as:

- Qualified sign language interpreters.
- Written information in other formats (large print, audio, accessible electronic formats, other formats).

Provides free language assistance services to people whose primary language is not English, which may include:

- Qualified interpreters.
- Information written in other languages.

If you need reasonable modifications, appropriate auxiliary aids and services, or language assistance services, contact the Nondiscrimination Grievance Coordinator at the address, phone number, fax number, or email address below.

If you believe that Aspirus Health Plan, Inc. has failed to provide these services or discriminated in another way on the basis of race, color, national origin, age, disability, or sex, you can file a *grievance* with:

Nondiscrimination Grievance Coordinator  
Aspirus Health Plan, Inc.  
PO Box 1890  
Southampton, PA 18966-9998  
Phone: 1-866-631-5404 (TTY: 711)  
Fax: 763-847-4010  
Email: customerservice@aspirushealthplan.com

You can file a *grievance* in person or by mail, fax, or email. If you need help filing a *grievance*, the Nondiscrimination Grievance Coordinator is available to help you.

You can also file a civil rights complaint with the U.S. Department of Health and Human Services, Office for Civil Rights, electronically through the Office for Civil Rights Complaint Portal, available at <https://ocrportal.hhs.gov/ocr/portal/lobby.jsf>, or by mail or phone at:

U.S. Department of Health and Human Services  
200 Independence Avenue, SW  
Room 509F, HHH Building  
Washington, D.C. 20201  
1.800.368.1019, 800.537.7697 (TDD)

Complaint forms are available at <http://www.hhs.gov/ocr/office/file/index.html>. This notice is available at Aspirus Health Plan, Inc.'s website: [https://aspirushealthplan.com/webdocs/70021-AHP-NonDiscrim\\_Lang-Assist-Notice.pdf](https://aspirushealthplan.com/webdocs/70021-AHP-NonDiscrim_Lang-Assist-Notice.pdf).

## Language Assistance Services

**Albanian:** KUJDES: Nëse flitni shqip, për ju ka në dispozicion shërbime të asistencës gjuhësore, pa pagesë. Telefononi në 1-800-332-6501 (TTY: 711).

**Arabic:** تنبيه: إذا كنت تتحدث اللغة العربية، فإن خدمات المساعدة اللغوية متاحة لك مجاناً. اتصل بن أعلى رقم الهاتف 1-800-332-6501 (رقم هاتف الصم والبك : 711)

**French:** ATTENTION: Si vous parlez français, des services d'aide linguistique vous sont proposés gratuitement. Appelez le 1-800-332-6501 (ATS: 711).

**German:** ACHTUNG: Wenn Sie Deutsch sprechen, stehen Ihnen kostenlos sprachliche Hilfsdienstleistungen zur Verfügung. Rufnummer: 1-800-332-6501 (TTY: 711).

**Hindi:** यान द : य द आप िहंदी बोलते ह तो आपके िलए मु त म भाषा सहायता सेवाएं उपल थ ह 1-800-332-6501 (TTY: 711) पर कॉल कर ।

**Hmong:** LUS CEEV: Yog tias koj hais lus Hmoob, cov kev pab txog lus, muaj kev pab dawb rau koj. Hu rau 1-800-332-6501 (TTY: 711).

**Korean:** 주의: 한국어를 사용하시는 경우, 언어 지원 서비스를 무료로 이용하실 수 있습니다. 1-800-332-6501 (TTY: 711) 번으로 전화해 주십시오.

**Polish:** UWAGA: Jeżeli mówisz po polsku, możesz skorzystać z bezpłatnej pomocy językowej. Zadzwoń pod numer 1-800-332-6501 (TTY: 711).

**Russian:** ВНИМАНИЕ: Если вы говорите на русском языке, то вам доступны бесплатные услуги перевода. Звоните 1-800-332-6501 (телетайп: 711).

**Spanish:** ATENCIÓN: si habla español, tiene a su disposición servicios gratuitos de asistencia lingüística. Llame al 1-800-332-6501 (TTY: 711).

**Tagalog:** PAUNAWA: Kung nagsasalita ka ng Tagalog, maaari kang gumamit ng mga serbisyo ng tulong sa wika nangwalang bayad. Tumawag sa 1-800-332-6501 (TTY: 711).

**Traditional Chinese:** 注意：如果您使用繁體中文，您可以免費獲得語言援助服務。請致電 1-800-332-6501 (TTY: 711)

**Vietnamese:** CHÚ Ý: Nếu bạn nói Tiếng Việt, có các dịch vụ hỗ trợ ngôn ngữ miễn phí dành cho bạn. Gọi số 1-800-332-6501 (TTY: 711).

**Pennsylvania Dutch:** Wann du Deutsch (Pennsylvania German / Dutch) schwetzscht, kannst du mitaue Koschte ebbergricke, ass dihr helft mit die englisch Schprooch. Ruf selli Nummer uff: Call 1-800-332-6501 (TTY: 711).

**Lao:** ໂປດຊາບ: ຖ້າວ່າທ່ານເວົ້າພາສາລາວ, ການບໍລິການຊ່ວຍເຫຼືອດ້ານພາສາໂດຍບໍ່ເສັຽຄ່າ, ຄວນມີພ້ອມໃຫ້ທ່ານ. ໂທ 1-800-332-6501 (TTY: 711).