N04202

Medical Policy-Genetic, Genotype and Genomic Testing

Values

Accountability ● Integrity ● Service Excellence ● Innovation ● Collaboration

Abstract Purpose:

The purpose of this policy to provide guidance for decisions related to genetic, genotype and genomic testing for Network Health Plan/Network Health Insurance Corporation/Network Health Administrative Services, LLC's (NHP/NHIC/NHAS) utilization management teams.

Policy Detail:

Refer to the appropriate Certificate of Coverage, Summary Plan Description, Individual and Family Policy, Evidence of Coverage or It's Your Choice booklet to determine eligibility, and coverage because Employer Group/Plan Sponsor and government contracts may vary. NHIC follows Medicare's National and Local (Wisconsin area) Coverage Determinations for its Medicare Advantage membership.

Procedure Detail:

I. Description
   a. "A genetic, genotype or genomic test is the analysis of human, viral, or tumor, DNA, RNA, chromosomes, proteins, or certain metabolites in order to detect alterations related to a heritable disorder or acquired disorder. This can be accomplished by directly examining the DNA or RNA that makes up a gene (direct testing), looking at markers co-inherited with a diseasecausing gene (linkage testing), assaying certain metabolites (biochemical testing), or examining the chromosomes (cytogenetic testing)." Genetic, genotype or genomic tests are conducted for a number of purposes, including but not limited to, predicting disease risk, newborn screening, determining clinical management, identifying carriers, and establishing prenatal or clinical diagnoses or prognoses in individuals, families, or populations.

II. Medical Indicators
   a. Genetic, genotype or genomic testing may be medically necessary when all of the following criteria are met:
      i. There must be a reasonable expectation based on family history, pedigree analysis, risk factors, and/or symptomatology that a genetically heritable or acquired condition exists.
         1. Rationale: Many genetic, genotype or genomic tests are imperfect predictors of either existing disease, disease susceptibility and/ or disease prognostic/therapeutic factors, particularly when used in the context of population screening, where individuals without family histories of disease, risk factors or symptoms are tested. For example, the probability exists that a disease may still occur, even when a negative/normal test result is obtained. Conversely, a specific disease may not occur when there is a positive/abnormal test result. While these concepts hold true for at-risk individuals as well, the probability of both these occurrences is greater in population screening, so test results are more difficult to interpret in a manner that will meaningfully

impact health outcomes. With a few limited exceptions (e.g., PKU testing), general screening of populations for diseases that can be attributed to genetic mutations is not advocated in the published scientific literature.

ii. The genotypes to be detected by a genetic, genotype or genomic test must be shown by scientifically valid methods to be associated with the occurrence of the disease, and the analytical and clinical validity of the test must be established. The observation must be independently replicated and subject to peer review.
    1. Rationale: Analytical Validity is an indicator of how well a test measures the property or characteristic it is intended to measure, and it is made up of three components:
        a. analytical sensitivity: the test is positive when the relevant gene mutation is present;
        b. analytical specificity: the test is negative when the gene mutation is absent; and
        c. reliability: the test obtains the same result each time
    2. Clinical Validity in genetic testing is a measurement of the accuracy with which a test identifies or predicts a clinical condition and involves the following:
        a. Clinical sensitivity: the probability that the test is positive if the individual being tested actually has the disease or a predisposition to the disease
        b. Clinical specificity: the probability that the test is negative if the individual does not have the disease or a predisposition to the disease
        c. Positive predictive value: the probability that an individual with positive test results will get the disease
        d. Negative predictive value: the probability that an individual with negative test results will not get the disease
        e. Heterogeneity: different mutations within the same gene may cause the same disease and can result in different degrees of disease severity; a failure to detect all disease-related mutations reduces a test's clinical sensitivity
        f. Penetrance: the probability that the disease will appear when a disease-related genotype is present. Penetrance is incomplete when other genetic or environmental factors must be present for a disease to develop.
        g. There are both benefits and risks associated with genetic, genotype or genomic tests. Genetic, genotype or genomic tests that are not fully assessed for analytical and clinical validity prior to their use in clinical practice have the potential for causing harm to patients. For example, patients who are wrongly classified as at-risk may be subjected to increased and unnecessary surveillance or treatments, some of which may be harmful, or even irreversible. Likewise, false negative test results may lead to delays in diagnosis and treatment.
    3. A probable therapeutic benefit of the genetic, genotype or genomic test must be established for the member/participant, i.e. test results will directly impact clinical decision making and/or clinical outcomes for the member/participant who is the subject of the test.
        a. Rationale: The development of genetic, genotype or genomic tests that can diagnose disease, predict disease occurrence, or determine disease prognostic/therapeutic factors has far outpaced the development of interventions to treat, improve or prevent those same diseases. Therapeutic benefit refers to the ability of genetic, genotype or genomic test results, either

positive or negative, to provide information that is of value in the clinical setting. Specifically for positive/abnormal test results, this could involve instituting treatments or surveillance measures, making decisions concerning future conception, or avoiding harmful treatments. Negative/normal test results can have therapeutic benefit in that unnecessary treatments or surveillance can be avoided. In the absence of such interventions, the benefits of testing are limited.

4. Genetic, genotype and/or genomic testing of children to predict adult onset diseases is not considered medically necessary unless direct medical benefit will accrue to the child, and in the case of adult onset disease, this benefit would be lost by waiting until the child has reached adulthood.
    a. Rationale: It is generally accepted in the published literature that unless useful medical intervention can be offered to children as a result of testing, formal testing should wait until the child is old enough to understand the consequences of testing and request it for him or herself. Ethical concerns related to the testing of children include the breach of confidentiality that is required by revealing test results to parents, the lack of ability to counsel the child in a meaningful way regarding the risks and benefits of testing, the impact a positive test could have in terms of discrimination, and the potential psychological damage that could occur from distorting a family's perception of the child.

b. Additional Information
    i. It may be necessary to offer pre-test genetic counseling from a qualified professional as well as post-test genetic counseling. This will be at the discretion of the provider and/or medical director.
        1. Rational:The consequences of genetic, genotype and/or genomic testinand the interpretation of test results is complex. Positive/abnormal test results do not necessarily mean that an individual will develop a specific disease. Conversely, negative/normal test results do not necessarily mean that an individual will not develop the disease for which testing is completed. Multiple mutations could exist for a single disease, some of which could be missed in testing. Not all mutations have the same effects in terms of disease severity. Some diseases may be caused by the interaction of both genetic and environmental factors. Effective treatments may not be available for some diseases; this is known as a "therapeutic gap." There may be significant emotional and psychological effects as a result of genetic, genotype and/or genomic testing. This complexity and the benefits and risks of genetic genotype and/or genomic testing should be fully disclosed to individuals prior to testing, just as counseling concerning the test results should be available.

III. Coverage
a. NHP/NHIC/NHAS may extend coverage of genetic, genotype or genomic testing for medically necessary indications as noted in this policy. NHIC follows CMS National Coverage Determinations (NCD) and Local Coverage Determinations (LCD) for application to its Medicare Advantage membership.

IV. Limitations/Exclusions
a. NHP/NHIC/NHAS considers genetic, genotype or genomic testing that does not meet the criteria as indicated not a covered service.

V. References

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Related Policies:

None

Related Documents:

None

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