Approved Revision: Do Not Implement Until 8/31/21

Whole Exome and Genome Sequencing

DESCRIPTION

Whole exome sequencing (WES) is targeted next-generation sequencing of the subset of the human genome that contains functionally important sequences of protein-coding DNA, while whole genome sequencing (WGS) uses next-generation sequencing techniques to sequence both coding and noncoding regions of the genome. WES and WGS have been proposed for use in individuals presenting with disorders and anomalies not explained by standard clinical workup.

Potential candidates for whole exome sequencing include individuals who present with complex genetic phenotypes suspected of having a rare genetic condition, who cannot be diagnosed by standard clinical workup, or when features suggest a broad differential diagnosis that would require evaluation by multiple genetic tests. The standard approach to the diagnostic evaluation of an individual suspected of having a rare genetic condition may include combinations of radiographic, biochemical, electrophysiological, and targeted genetic testing such as a chromosomal microarray, single-gene analysis, and/or a targeted gene panel. Identifying a molecularly confirmed diagnosis in a timely manner for an individual with a rare genetic condition can have a variety of health outcomes, including guiding prognosis and improving clinical decision-making. Testing the affected individual and both parents (trio testing) is recommended to increase the chance of finding a definitive diagnosis and reduce false-positive findings. Testing of one available parent should be done if both are not immediately available and one or both parents can be done later if needed.

POLICY

- Whole exome sequencing (WES) may be considered ***medically necessary*** if the medical appropriateness criteria are met **(See Medical Appropriateness below)**.

- Whole genome sequencing (WGS) is considered ***investigational.***

- Pre- and post- genetic counseling as an adjunct to genetic testing is considered ***medically necessary***.

MEDICAL APPROPRIATENESS

- Whole exome sequencing (WES) may be considered **medically appropriate** if **ALL** of the following are met:

    - Testing is indicated for **One or More** of the following:
        - Diagnostic testing of individual 18 years of age or younger
        - Screening of parent(s)
    - Congenital anomalies and/or neurodevelopmental disorder(s) cannot be explained by standard non-invasive tests
    - There is potential for change in treatment of the affected individual
    - Documentation of prior genetic testing as **ANY ONE** of the following:
        - Previous phenotype genetic testing (e.g., targeted single gene or chromosomal microarray analysis) was negative and genetic etiology is the most likely explanation
        - Previous genetic testing (e.g., targeted single gene or chromosomal microarray analysis) has failed to yield a diagnosis and invasive procedures or testing is the next diagnostic step (e.g., muscle biopsy)

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IMPORTANT REMINDERS

- Any specific products referenced in this policy are just examples and are intended for illustrative purposes only. It is not intended to be a recommendation of one product over another and is not intended to represent a complete listing of all products available. These examples are contained in the parenthetical e.g. statement.

- We develop Medical Policies to provide guidance to Members and Providers. This Medical Policy relates only to the services or supplies described in it. The existence of a Medical Policy is not an authorization, certification, explanation of benefits or a contract for the service (or supply) that is referenced in the Medical Policy. For a determination of the benefits that a Member is entitled to receive under his or her health plan, the Member's health plan must be reviewed. If there is a conflict between the Medical Policy and a health plan, the express terms of the health plan will govern.

ADDITIONAL INFORMATION

Published evidence of well-designed studies in peer review journals is insufficient to determine whether the use of whole genome sequencing tests improve health outcomes.

SOURCES

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