Quartz

Whole Exome and Whole Genome Sequencing

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Policy

The Medical Management Department reviews referral requests for authorization of whole exome and whole genome sequencing.

This Medical Policy does not constitute medical advice. When deciding coverage, the enrollee's specific plan document must be referenced. The terms of an enrollee's plan document (Certificate of Coverage [COC] or Summary Plan Description [SPD]) may differ from this Medical Policy. In the event of a conflict, the enrollee's specific benefit plan document supersedes this Medical Policy. All reviewers must first identify enrollee eligibility, any federal or state regulatory requirements, and the plan benefit coverage prior to use of this Medical Policy. Other Policies and Coverage Determination Guidelines may apply. Quartz reserves the right, in its sole discretion, to modify its Policies and Guidelines as necessary.

Procedure

A. Documentation Requirements:

In order to facilitate the authorization process, referral requests must include the following:

1. Documentation that the patient has been evaluated by a board-certified/eligible medical geneticist or other board-certified/eligible specialist physician with a specific expertise in the (genetic) conditions which are considered likely.
2. Order by a board-certified/eligible medical geneticist or genetic qualified nurse or pediatrician, neurologist or psychiatrist in coordination with genetics.
3. Documentation that pretest counseling has been performed and posttest counseling is planned.

B. Criteria for Medical Necessity

Whole exome sequencing is medically necessary if **ALL** of the following are met:

1. A genetic disorder is likely to be the cause of the symptoms/abnormalities as displayed by **ONE** of the following:
    a. Presence of multiple abnormalities affecting unrelated organ systems; **OR**
    b. **TWO** of the following:
        i. Abnormality in at least one organ system;
        ii. Family history is strongly suggestive of a genetic etiology;
        iii. Significant intellectual disability, complex neurodevelopmental disorder or severe neuropsychiatric condition;
        iv. Unexplained developmental regression;
        v. Infant with biochemical findings suggestive of an inborn error of metabolism or complex metabolic phenotype (e.g, high ammonia levels or lactic acidosis); **AND**

2. No other causative circumstances exist to explain the symptoms/abnormalities, (e.g., infection, injury, environmental exposure); **AND**
3. Symptoms/abnormalities do not suggest a condition for which single or targeted gene testing is available **OR** such testing has been performed and is negative; **AND**
4. A diagnosis cannot be made by standard clinical work-up including single gene mutation testing for specific conditions and/or testing may preclude the need for invasive procedures for diagnosis (e.g., biopsy or invasive testing); **AND**
5. Testing is predicted to have an impact on health outcomes through **ONE** of the following:
    a. Determining prognosis or appropriate treatment plan; **OR**
    b. Avoidance of invasive testing for diagnostic purposes; **OR**
    c. Avoidance of future testing for screening purposes if such testing could be avoided through the results of WES.

Note: Testing of the biological mother and father of the person undergoing testing (i.e., family trio/comparative testing) is considered medically necessary when criteria are met for this person to be tested and is performed concurrently, or this person's testing has been previously performed.

C. Indications Considered Experimental and Investigational (Not an all-inclusive list):

1. Whole exome sequencing or whole genome sequencing for tumor mutations / cancer testing.
2. Whole genome testing for any indication including newborn screening.
3. Whole mitochondrial genome sequencing for any indication.
4. Whole exome sequencing of fetus during pregnancy or a terminated fetus.

References:

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