Genetic Testing Guidelines effective on July 25, 2025

Chromosomal Microarray Analysis

- Postnatal/Pediatric evaluation - Expansive edit to include neonatal death to the list of indications considered medically necessary for chromosomal microarray analysis. (coverage expansive)
- New section for Optical Genome Mapping clarifies current position as not medically necessary. OGM may be an alternative methodology for structural variant analysis, but more studies are required before considering this technique as medically necessary. (content clarification)

Whole Exome Sequencing and Whole Genome Sequencing

- Clarify and restructure the criteria for improved readability. (content clarification)
- Restrictive edit specifies that WES for early neonatal death is an exclusion. (restrictive)

Pharmacogenomic Testing

- For each of the therapies and associated biomarkers in Table 1, genotyping for the appropriate biomarker is considered medically necessary when ALL the following conditions are met – (clarifications)
- Clarified title of Table - Table 1. Therapies and associated biomarkers considered medically necessary for genotyping (clarification)
- "Expansive changes:
    - Donanemab-azbt added for association with genotyping for ApoE ε4 in the realm of Neurology for treatment of Alzheimer's disease
    - Deuruxolitinib added for association with genotyping for CYP2C9 in the realm of Dermatology for treatment of alopecia areata
    - NUDT15 risk allele added to explain the majority of thiopurine-related myelosuppression risk in Asians and Hispanics.
- Clarification: eliglustat's therapeutic area clarified as being related to hematology rather than pediatrics

Predictive and Prognostic Polygenic Testing

- Guideline reaffirmed. Edited Description/Scope and Rationale.