NGS Panel Expand

**Test Code:** MMUPG
**Turnaround time:** 4 weeks
**CPT Codes:** 81479 x1

### Condition Description

**What is the NGS Panel Expand?**

The NGS Panel Expand (NGSExpand) includes analysis of approximately 5,000 genes associated or implicated with Mendelian disease. This analysis utilizes previously unseen data from a completed NGS panel performed at EGL Genetics and incorporates information regarding a patient's clinical presentation to help identify variants related to the patient's phenotype.

An eligible NGS panel must have been previously performed at EGL Genetics in order to request this test. Eligible panels include most NGS panels performed January 2016 or later; sponsored testing does not apply. Please contact the laboratory with any questions regarding panel eligibility.

**Is the NGSExpand appropriate for my patient?**

This testing is appropriate for patients with a previously negative or non-diagnostic NGS panel performed at EGL Genetics where a genetic diagnosis is still suspected. Common scenarios for the NGSExpand include:

- the patient has a complex phenotype (example: patient has several features that overlap with more than one syndrome);
- previous testing did not include all of the common genes associated with the patient's clinical presentation (example: patient presented with muscle weakness and had a negative Limb Girdle Muscular Dystrophy Panel);
- the patient has developed additional clinical features since the time of previous testing (example: a patient who presented with seizures and had a negative epilepsy panel now has appreciable dysmorphic features).

**What is needed to order the NGSExpand?**

Since this analysis uses existing data, no new sample is needed to initiate testing. To order the NGSExpand, fax or email a completed requisition along with recent clinic notes and a consent form to EGL Genetics.

**What may be reported as part of the NGSExpand?**

The NGSExpand reports variants in genes that are considered to be related to or possibly related to the patient's phenotype. This analysis may reveal variants in genes that are not associated with the patient's phenotype, including information regarding carrier status or adult-onset disorders. A consent form is required to receive information not related to the patient's phenotype.

For more information on the NGSExpand, please contact EGL Genetics at 470-378-2200.

### Indications

The NGES Expand is indicated when there is a suspicion of a genetic etiology contributing to the proband’s manifestations. This testing is appropriate for patients with a previously negative or non-diagnostic NGS panel performed at EGL Genetics where a genetic diagnosis is still suspected.

### Methodology

NGS Panel Expand is performed by re-analyzing raw NGS sequencing data generated from this individual's previous NGS panel performed at EGL genetics. This re-analysis includes 5,089 genes implicated in disease. The target region includes the exon and 10bp of flanking intronic sequence. The DNA sequence is mapped to and analyzed in comparison with the published human genome build UCSC hg19 reference sequence. The targeted coding exons and splice junctions of the known protein-coding RefSeq genes are assessed for the depth of coverage and data quality threshold values. Variants with 15X or more coverage are analyzed. EGL has developed an NGS Panel Upgrade bioinformatics analysis pipeline to compare sequence changes in the individual being tested to the reference sequence. High-quality single nucleotide variants (SNVs) which pass EGL's quality filters are not confirmed by Sanger sequencing. Reportable SNVs that do not pass the quality filters are confirmed using bidirectional Sanger sequence analysis.

### Detection

Unknown.

### Specimen Requirements

Submit only 1 of the following specimen types

### Special Instructions

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Please submit medical records or clinical summary notes, and a signed consent form when ordering NGSExpand. Please use the requisition form below

NGS Panel Expand Requisition form