



August 7, 2018

Seema Verma, CMS Administrator  
Centers for Medicare & Medicaid Services  
7500 Security Boulevard  
Baltimore, MD 21244

Re: 2018 Preliminary Gapfill for Codes 81425, 81426, and 81427

Dear Administrator Verma:

The American College of Medical Genetics and Genomics (ACMG) welcomes the opportunity to comment on the 2018 CLFS Gapfill Preliminary Determinations. ACMG is the only nationally recognized professional membership organization dedicated to improving health through the practice of medical genetics and genomics. Our membership includes over 2000 genetics professionals, nearly 80% of which are board certified clinical and laboratory geneticists and genetic counselors.

ACMG has significant concerns about the preliminary gapfill determinations for the whole genome sequencing codes 81425, 81426, and 81427. We believe that CMS MACs lacked adequate data to support a gapfill process for these codes. The gapfill process lacks transparency, but it is clear that the preliminary determinations do not factor in actual costs. These include analytical components of sequencing, staff labor, and reagents for which limited cost data suggests a combined cost of approximately \$1,800 with interpretive costs of bioinformatics and expert interpretation being an additional \$1,800. This totals \$3,600 but excludes data storage, overhead, and other costs that are not accounted for in our limited cost data. The result of relying on inadequate data is a set of price recommendations that are significantly below the actual cost of providing these services. Further, there are other genomic sequencing codes priced on the CLFS that could serve as viable crosswalks for these testing services. Specifically, we provide the following crosswalk recommendations for the whole genome sequencing codes 81425, 81426, and 81427.

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Executive Director

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7101 Wisconsin Avenue  
Suite 1101, Bethesda, MD 20814  
Telephone 301-718-9603  
Fax 301-718-9604  
[www.acmg.net](http://www.acmg.net)

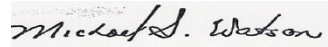
<b>CPT Code</b>	<b>Code Description</b>	<b>Test Purpose and Method</b>	<b>Crosswalk Recommendation</b>
81425	Genome (e.g., unexplained constitutional or heritable disorder or syndrome); sequence analysis	Used to detect the genetic basis of unexplained heritable disorders or syndrome in coding and noncoding regions of the genome, including detection of CNVs, structural rearrangements, and intergenic variants/events	81415
81426	Genome (e.g., unexplained constitutional or heritable disorder or syndrome); sequence analysis, each comparator genome (e.g., parents, siblings) (List separately in addition to code for primary procedure)	Used as the comparator for 81425	81415
81427	Genome (e.g., unexplained constitutional or heritable disorder or syndrome); re-evaluation of previously obtained genome sequence (e.g., updated knowledge or unrelated condition/syndrome)	Used to reinterpret previously obtained sequence data considering novel medical information or changes/development of clinical phenotype; performed using extensive bioinformatics analysis and professional review	81417

Although the current cost of whole genome sequencing is higher than whole exome sequencing, we believe that the crosswalk recommendations above are justified based on similarities in the technology and expert interpretation required for both types of sequencing. While there are differences between the two methodologies, both rely on massively parallel sequencing, are not limited to a subset of genes, require extensive bioinformatics, and require complex interpretation of results by highly trained professionals.

Unlike whole exome sequencing which covers about 1% of the human genome, whole genome sequencing covers both the coding and noncoding regions of the genome and can detect large copy number variations, structural rearrangements, and insertions/deletions. While the cost of sequencing itself may come nearer to that of whole exome sequencing in the future, the total cost is expected to remain higher, especially when considering costs associated with complex bioinformatics, expert interpretation, and data storage.

In summary, gapfill is not an appropriate method for determining the price for whole genome sequencing codes 81425, 81426, and 81427. We strongly encourage CMS to reevaluate the approach for determining the price for these codes. The preliminary prices determined by gapfill are unrealistically low and would cause significant financial burden on laboratories that offer these services. We hope the information above will help CMS identify a more appropriate approach for pricing whole genome sequencing services.

Sincerely yours,

A handwritten signature in black ink that reads "Michael S. Watson". The signature is written in a cursive style and is placed on a light gray rectangular background.

Michael S. Watson, MS, PhD, FACMG  
Executive Director