Reproductive Testing: Carrier Screening 2025.2

Effective: 07/01/2025 Last Revision: 02/18/2025 Last Clinical Review: 01/31/2025

Expanded Carrier Screening Panels

- I. Expanded carrier screening panels¹ may be considered **medically necessary** when:
 - A. The member is considering pregnancy or is currently pregnant², **AND**
 - B. The panel includes the genes CFTR and SMN1.
- II. Expanded carrier screening panels are considered **investigational** for all other indications.

¹ Fragile X (81243) and spinal muscular atrophy (SMA) (81329) carrier screening may be billed along with 81443 if performed separately from the remainder of the panel per CPT Code Book Guidelines. If 81243 is billed along with 81443, the member should still meet the specific Fragile X syndrome criteria.

² ACMG recommends follow-up screening for the partner of the member that is pregnant or considering pregnancy via analysis of the same gene that has the pathogenic or LP variant as identified in the member. Therefore, expanded carrier screening panels are not recommended to be completed by both reproductive partners in tandem.

REFERENCES

- Committee Opinion No. 690 (Reaffirmed 2023: Carrier Screening in the Age of Genomic Medicine. *Obstet Gynecol*. 2017;129(3):e35-e40. doi:10.1097/AOG.00000000001951
- Gregg AR, Aarabi M, Klugman S, et al. Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG) [published online ahead of print, 2021 Jul 20] [published correction appears in Genet Med. 2021 Aug 27;:]. Genet Med. 2021;10.1038/s41436-021-01203-z. doi:10.1038/s41436-021-01203-z

