



Subject: Preconception Genetic Carrier Screening*

Effective Date: April 30, 2014

Department(s): Utilization Management

Policy: Preconception genetic carrier screening is reimbursable under Plans administered by QualCare, Inc. The scope of testing considered medically necessary as supported by professional consensus and current literature is outlined in the **Procedure** section below.

Objective: To ensure proper and consistent reimbursement and to limit coverage to treatment that is shown to have scientific validity.

Procedure: A. For individuals of Ashkenazi Jewish descent, testing for genetic carrier status of the following disorders is considered medically necessary:

- Bloom Syndrome(**CPT 81209**)
- Canavan Disease (**CPT 81200**)
- Cystic Fibrosis (**CPT 81220**)
- Familial Dysautonomia (**CPT 81260**)
- Fanconi Anemia (group C) (**CPT 81242**)
- Gaucher Disease (**CPT 81251**)
- Mucopolidosis IV (**CPT 81290**)
- Neiman-Pick Disease (type A) (**CPT 81330**)
- Tay-Sachs Disease (**CPT 81255**)

B. For individuals of African, Southeast Asian and Mediterranean descent, testing for the carrier status of the

following disorders is considered medically necessary:

- Hemoglobinopathies (sickle cell disease, thalassemias) (CPT 81401, 81257)

C. Panethnic carrier screening is considered medically necessary for cystic fibrosis(using the standard 25 mutation panel) (**CPT 81220**) and spinal muscular atrophy(**CPT 81401**).

D. Carrier testing for other specific genetic diseases is considered medically necessary if genetic counseling shows that the couple has risk of the heritable condition and the condition's natural history has high morbidity, the carrier state cannot be diagnosed by alternate tests, and the genetic test has established adequate performance characteristics(carrier detection rate $\geq 95\%$). Examples include Rett Syndrome, Long QT Syndrome, Huntington Disease and fragile X Syndrome.

E. Expanded carrier screening panels that use next-generation sequencing technology to screen for mutations across many genes are not considered medically necessary. There are multiple such test panels available on the market, each offering a different combination and total quantity of mutations tested for , with variable overlap among them. There is no current professional consensus or guidance regarding which disease genes or mutations to include. There is insufficient peer-reviewed data on the clinical validity and utility of these test panels.

Examples of currently marketed expanded screening panels include Counsyl™, GoodStart Select™, Inherigen™, Inheritest™ and Natera One™.

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Drafted By/Date: MMcNeil, MD 04/30/14

Approved By/Date: QM Committee 5/13/14

Revised By/Date: M. McNeil, MD 10/04/16

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Reviewed w/o Revision By/Date: MMcNeil, MD 06/25/18

Approved By/Date: QM Committee 08/21/18