Overview
The purpose of this document is to describe the guidelines Neighborhood Health Plan (NHP) utilizes to determine medical appropriateness for preimplantation genetic testing (PGT) which includes preimplantation genetic diagnosis (PGD) for single gene defects and translocations, and preimplantation genetic screening (PGS) for aneuploidy.

Coverage Guidelines
NHP covers medically necessary PGD for a debilitating, genetically-defined and genetically predictable disease with early onset mortality or morbidity when there is no known treatment for the condition or the available interventions are either inadequately effective or significantly burdensome. NHP does not cover PGS.

The specialist and/or the primary care provider are responsible for providing all necessary clinical information including medical history of patient and partner or child, where appropriate. It is expected that the member/couple are counselled regarding the testing alternatives to PGD (e.g. amniocentesis, chorionic villous sampling), potential risks of PGD (embryo arrest, diagnostic uncertainty and unknown long-term effects of PGD), and that traditional prenatal diagnostic testing may still be recommended after successful PGD and pregnancy.

Authorization of PGD is limited to the following criteria:

Preimplantation Genetic Diagnosis (PGD)
1. NHP covers medically necessary PGD to test for unbalanced chromosome rearrangements when one of the genetic parents is known to have a balanced reciprocal translocation or Robertsonian translocation, or to have a microdeletion /duplication with one previously affected child. NHP may require laboratory documentation of the genetic tests.

2. NHP covers medically necessary PGD to detect evidence of any of the following genetic disorders in an embryo when:
   a. Both genetic parents are known carriers of a single gene autosomal recessive disorder, or one of the genetic parents is a known carrier and they have a child who has been diagnosed with the disorder such as, but not limited to the following:
      i. Canavan disease
      ii. Cystic Fibrosis
      iii. Epidermolysis Bullosa Simplex (autosomal recessive type)
      iv. Familial dysautonomia
      v. Fanconi’s Anemia
      vi. Gaucher Disease

* Not all commercial plans cover this service, please check plan’s benefit package to verify coverage.
vii. Hurler Syndrome  
viii. Methylmalonic acidemia  
ix. Propionic academia  
x. Sickle Cell Anemia  
xi. Spinal Muscular Atrophy Type I  
xii. Spinocerebellar Ataxia (autosomal recessive type)  
xiii. Tay-Sachs Disease  
xiv. Thalassemia Syndromes  

b. One genetic parent is a known carrier of a single gene autosomal dominant disorder such as, but not limited to, the following:  
   i. Epidermolysis Bullosa (autosomal dominant type)  
   ii. Huntington's Disease  
   iii. Myotonic Dystrophy  
   iv. Neurofibromatosis Type II  
   v. Spinocerebellar Ataxia (autosomal dominant type)  
   vi. Tuberous sclerosis  

c. The genetic female parent is a known carrier of a single gene X-linked recessive disorder such as, but not limited to the following:  
   i. Adrenoleukodystrophy  
   ii. Alport Syndrome  
   iii. Becker muscular dystrophy  
   iv. Fabry disease  
   v. Choroideremia  
   vi. Duchenne muscular dystrophy  
   vii. Fragile X syndrome  
   viii. Hemophilia A & B  
   ix. Hunter Syndrome  
   x. Incontinentia pigmenti  
   xi. Lesch-Nyhan Syndrome  
   xii. X-linked intellectual disability  

Exclusions  
1. PGD services for any reason other than listed above.  
2. In-vitro fertilization or other assistive reproductive technologies when performed solely to accomplish PGT and when the listed criteria listed in the Infertility Services policy are not met.  
3. PGT as an adjunct to infertility services for members who are not eligible for such services as determined by the Preimplantation Genetic Testing clinical coverage criteria.  
4. PGD Services if the member or member’s spouse are using illicit substances or abusing substances known to negatively interfere with fertility or fetal development (e.g., marijuana, opiates, cocaine, or alcohol);  
5. PGT for:  
   a. Screening for aneuploidy including in the setting of: recurrent miscarriage, repeated failed implantation during IVF, or advanced maternal age.  
   b. Screening for genetic or chromosomal abnormalities or traits in the absence of a known, clinically significant genetic or chromosomal condition in the genetic parents.  
   c. Carrier testing to determine embryo’s carrier status.  
   d. Human Leukocyte antigen (HLA) typing of an embryo to identify a future suitable stem cell, tissue or organ transplantation donor.  
   e. Translocations which will always produce an abnormal gamete such as 45XX (21;21) & 45XY(21;21)  
   f. Gender selection in the absence of a documented X-linked disorder.  
   g. Selecting non-medical traits.
h. Selecting against predisposition to disease when there is no single known genetic or chromosomal defect that definitively causes the disease.
i. Late onset/adult onset disorders that are not listed in criteria above.
j. Genetic conditions contributed to by donor egg and sperm.

Definitions
Autosomal Dominant: Autosomal dominant is one of several ways that a trait or disorder can be passed down through families. If a disease is autosomal dominant, it means you only need to get the abnormal gene from one parent in order for you to inherit the disease. One of the parents may often have the disease.

Autosomal Recessive: A disorder characterized by two mutated copies of the gene must be present in each cell in order for the disease or trait to develop. Affected persons usually have two unaffected parents who each carry a single copy of the mutated gene and they are known as carriers.

Preimplantation Genetic Diagnosis: A test involving an embryo that has been created using assisted reproductive technology such as in-vitro fertilization. After the eggs are removed the eggs are fertilized. Those eggs which are successfully fertilized are developed into a blastomere or a blastocyst. One or two cells are removed in order to test for the specific genetic condition in question.

X-linked Dominant Disorders: Caused by mutations in the gene on the X chromosome. Females are more frequently affected than males and the chances of passing on an X linked dominant disorder differ between men and women.

X-Linked Recessive: Are caused by mutations in the genes on the X chromosome. Males are more frequently affected than females and the chances of passing on the disorder differ between men and women. Families with an X linked recessive disorder often have affected males but rarely affected females in each generation. A characteristic of X linked inheritance is that fathers cannot pass X-linked traits to their sons.

Related Policies
- Infertility Services

Effective
July 2017: Annual update. Added the exclusion “PGD Services if the member or member’s spouse are using illicit substances or abusing substances known to negatively interfere with fertility or fetal development (e.g., marijuana, opiates, cocaine, or alcohol)“
July 2016: Annual update
July 2015: Updated references
August 2014: Added language to Coverage Guidelines, “a debilitating genetic disease with early onset mortality or morbidity and when there is no known treatment for the condition or the available interventions are either inadequately effective or significantly burdensome.“ Added exclusions: 3f selecting for non-medical traits and 3j Genetic conditions contributed to by donor egg and sperm.
June 2013: Annual update, added specific genetic disorders.
June 2012: Effective date.

References

doi: 10.1136/bmj.g761


