Samples Selection Guide for PG-seq™ Core Panel

In this document we will describe the type and number of family samples required to obtain conclusive results in the PG-Seq™ Core Panel Analysis.

GENERAL POINTS

Generally, samples from the couple (progenitors of the embryo) and another additional family member for each mutation tested are required. It is recommended that all family members included were confirmed genetic carriers of the alteration. If they are not carriers they could also be included in the study, but its state should be completely known.

A healthy (non-carrier)/affected child is considered a family member, and he/she is the best option for obtaining successful results. If the child is only carrier (in recessive disorders), the origin of the alteration is always required to be known. If it is not possible, the sample should be discarded and a different sample from another family member should be used. If there are no child samples available, it is possible to perform the study with samples from other family members. In these cases, there are differences between recessive and dominant diseases.

AUTOSOMAL DOMINANT DISORDERS

In the case of disorders with dominant inheritance pattern, samples from the couple and a minimum of one sample from other affected family members are required (figure 1). For example, required sample from a grandparent (affected), and recommended from both grandparents, always in the affected family. Please note that, if the available sample comes from an affected member, the solution is clear. However, if the available sample is from a healthy individual, and we have no clinical confirmation that the other member (non-available) was a carrier, the risk can be high.

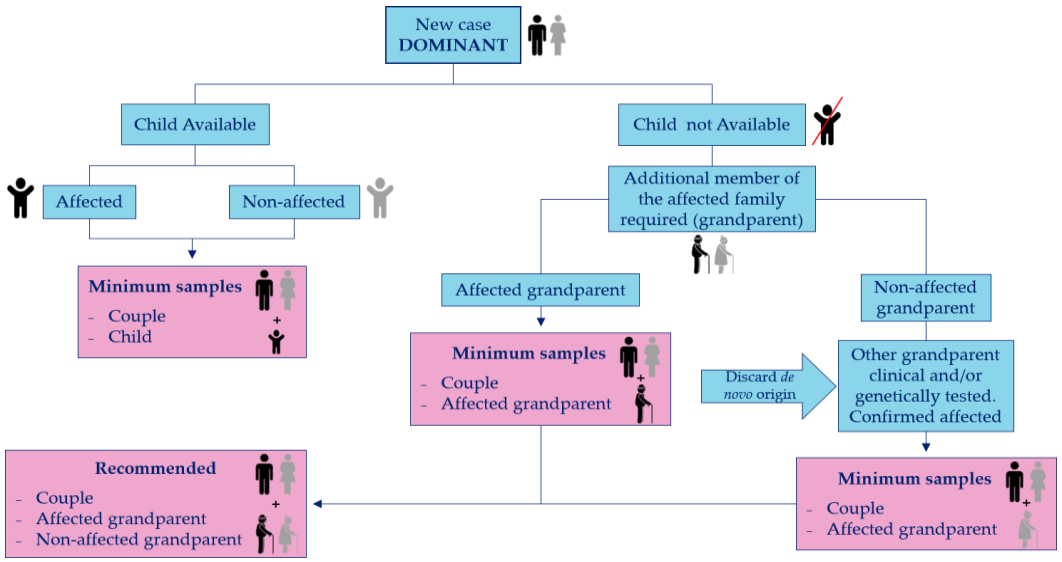

Figure 1. Diagram representing the minimum and recommended samples for a case of autosomal dominant disease (affected male).

AUTOSOMAL RECESSIVE DISORDERS

In disorders with recessive inheritance pattern, we will need more samples. We will need a minimum of one sample from each branch of the family (figure 2). Also, we need to be sure that, if only one member is available and the male/female-father/mother available is a non-carrier, the other family member may be genetically confirmed as a carrier.

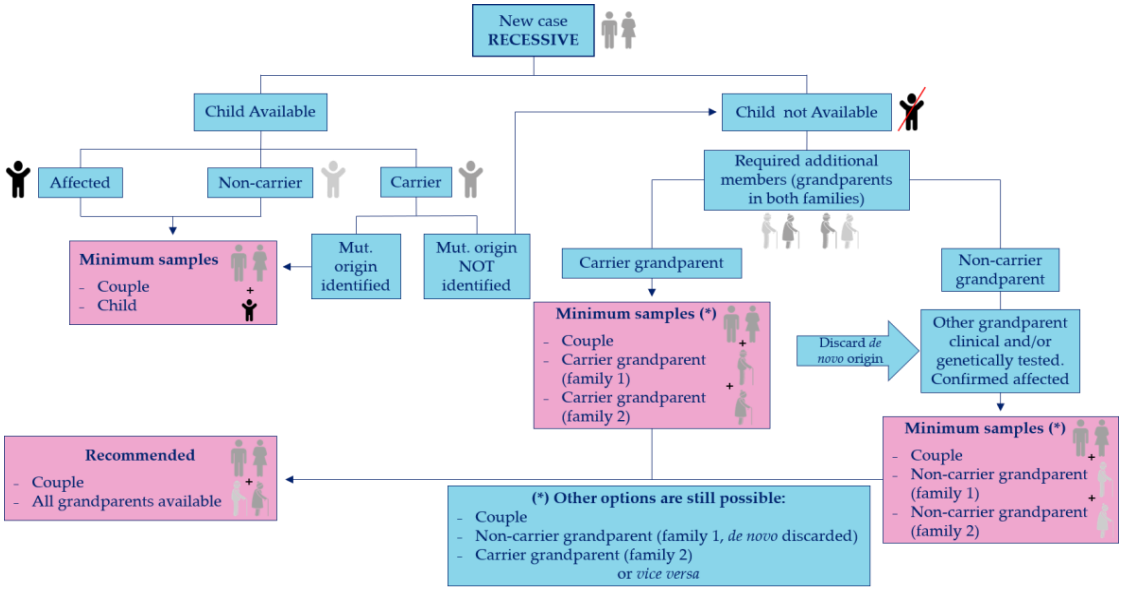

Figure 2. Diagram representing the minimum and recommended samples for a case of autosomal recessive disease (carrier male and female).

X-LINKED DISORDERS

In X-linked disorders, in a female carrier, we need samples from the female family branch, like in the case of dominant disorders (figure 3).

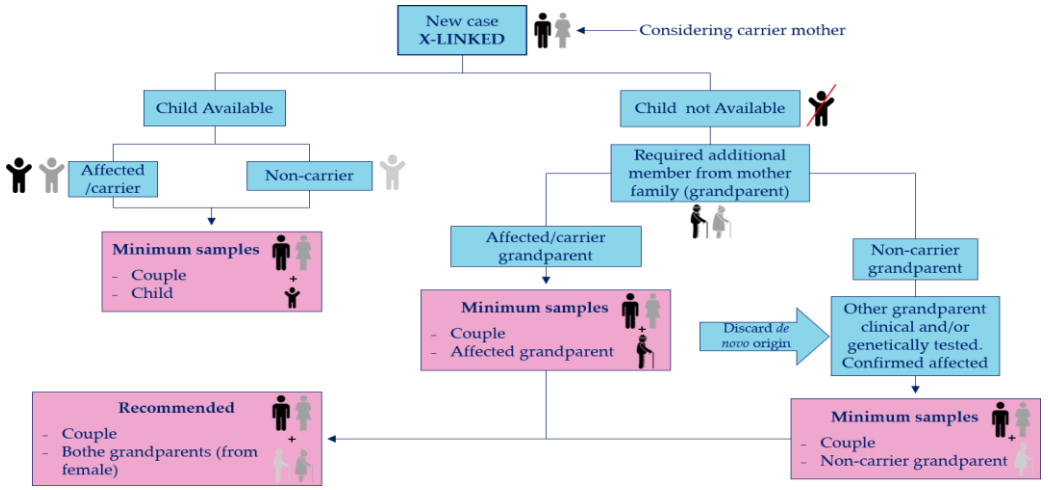

Figure 3. Diagram representing the minimum and recommended samples for a case of X-linked disorder (carrier female and affected male).