

PGT-A Preimplantation Genetic Testing for Aneuploidy



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Euploid/Aneuploid:

Samples reported as **euploid** have been found to be chromosomally normal within the limitations of Preimplantation Genetic Testing for Aneuploidy (PGT-A). Samples reported as **aneuploid** have been found to contain at least one aneuploid chromosome and is an abnormal result.

Gain and Loss:

Segmental aneuploidy indicates the presence of an extra or missing region of a chromosome respectively, but does not involve the entire chromosome. This is considered an abnormal result.

Complex Abnormal:

The diagnosis of 'complex abnormal' refers to embryo biopsy samples that demonstrate three or more chromosomal aneuploidies.



Mosaicism:

The term 'mosaicism' describes the occurrence of two or more genetically distinct populations of cells within an embryo biopsy sample. 'High-level' denotes an estimated level of mosaicism 50-80%, and 'Low level' denotes an estimated level of mosaicism of 20-50%. Levels of mosaicism estimated to be lower than 20% are not reported as they are not considered to be significant. Complex mosaic denotes three or more high/low level mosaicism occurrences. This reporting structure is in keeping with published international guidelines.

No Result:

Individual samples reported as 'No Result' did not yield a result, due to one of the following possible reasons:

1. The absence of intact DNA in the biopsy tube for analysis
2. The failure to amplify DNA within the biopsy sample due to inadequate sample quality
3. An individual sample that amplified adequately but which did not meet assay quality control criteria, consequently the data could not be interpreted

Sex Chromosomes:

Next Biosciences PGT-A reports include information on aneuploidy and copy-number variation for all chromosomes including X and Y. In some countries (e.g. South Africa*) it is unlawful to use this information for social sex selection. It is the responsibility of the ordering physician to use these data in accordance with applicable laws in their jurisdiction. [*South African Government Gazette 501: 2 March 2012, #35099, page 14, Item 13].