Version Date 02/01/2024

Smart PGT-A PLUS
Preimplantation Genetic Testing for Aneuploidies
by Igenomix

RESULTS GUIDE

PGT FOR ANEUPLOIDY SCREENING

Smart PGT-A

Smart PGT-A Plus

Igenomix®
PART OF VITROLIFE GROUP

This result guide is intended to provide an overview of your PGT-A results and is based on current knowledge and understanding, which is subject to refinement and change. It includes information pertaining to Igenomix's two PGT-A platforms, Smart PGT-A and Smart PGT-A Plus. This guide is not intended to replace genetic counseling. If you still have questions as to what your results mean after reading through this guide and reviewing the results with your providers, you are welcome to schedule an appointment with a genetic counselor at Igenomix. Please note that embryos are frozen and kept at the fertility clinic, and any decisions regarding embryo transfer, discard, or storage are made with the physician.

Contact us at:
Genetic Counseling department: (785) 485-0014, gc@igenomix.com
Igenomix Customer Support: (786) 401-7546, infousa@igenomix.com

TABLE OF CONTENTS

PGT-A BASICS

PGT FOR ANEUPLOIDY SCREENING (PGT-A)

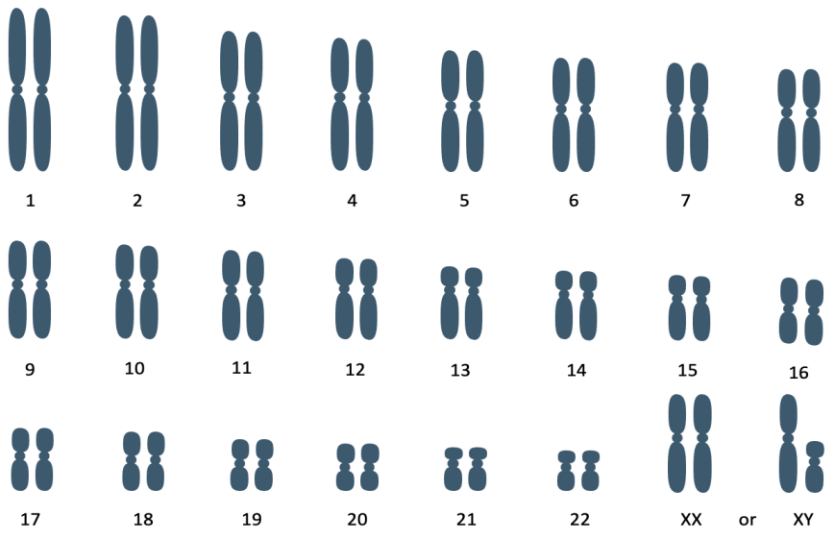
PGT-A BASICS

Preimplantation genetic testing for aneuploidy (PGT-A) is a genetic test performed on embryos to screen for numeric chromosome abnormalities (aneuploidy).

WHAT ARE CHROMOSOMES?

Chromosomes are the structures which store our genetic material. If you think of each gene as a book of instructions for how our bodies grow and develop, then a chromosome is like a bookshelf. Each chromosome stores many different genes.

Humans have 23 pairs of chromosomes (46 in total). The chromosomes are numbered 1 through 22, with the 23rd pair being the "sex chromosomes" (X and Y chromosomes). Below is an image of an example "karyotype" showing the typical number of chromosomes. Of note, a complete karyotype cannot be obtained by PGT-A; instead, next-generation sequencing (NGS) technology is used to detect changes in the number of chromosomes compared to the typical number, or so-called "chromosomal copy number variations."

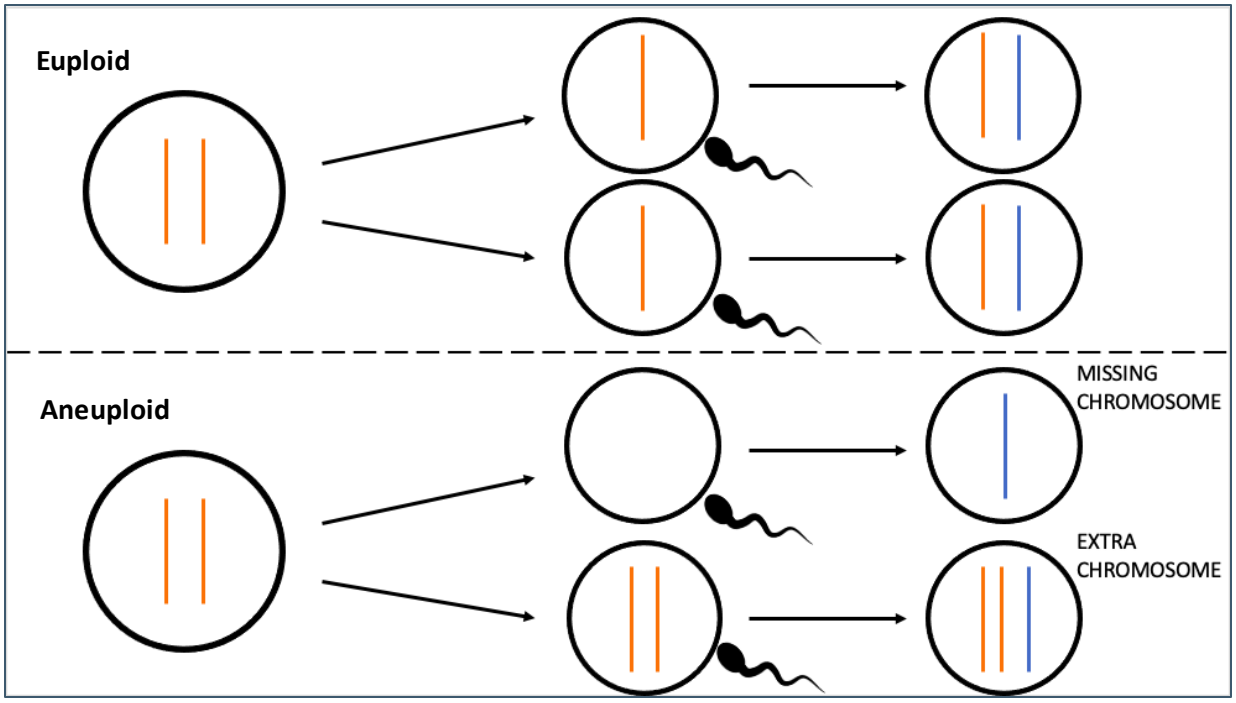


PGT-A BASICS (continued)

WHAT IS ANEUPLOIDY?

When an embryo has the typical number of chromosomes, it is called **euploid**. When an embryo does not have the typical number of chromosomes, it is called **aneuploid.** Embryos with chromosomal copy number changes detected by NGS, signifying an extra or missing chromosome, are considered to be aneuploid.

The egg and sperm typically have one copy of each chromosome. When the egg is fertilized by the sperm, it results in an embryo with **two copies of every chromosome**. Sometimes, during sperm or egg formation, a pair of chromosomes may become stuck together. This can result in one egg/sperm with an extra chromosome, and one egg/ sperm with a missing chromosome (see diagram below). Once fertilized, this can result in an embryo with an extra or missing chromosome (aneuploid embryo).



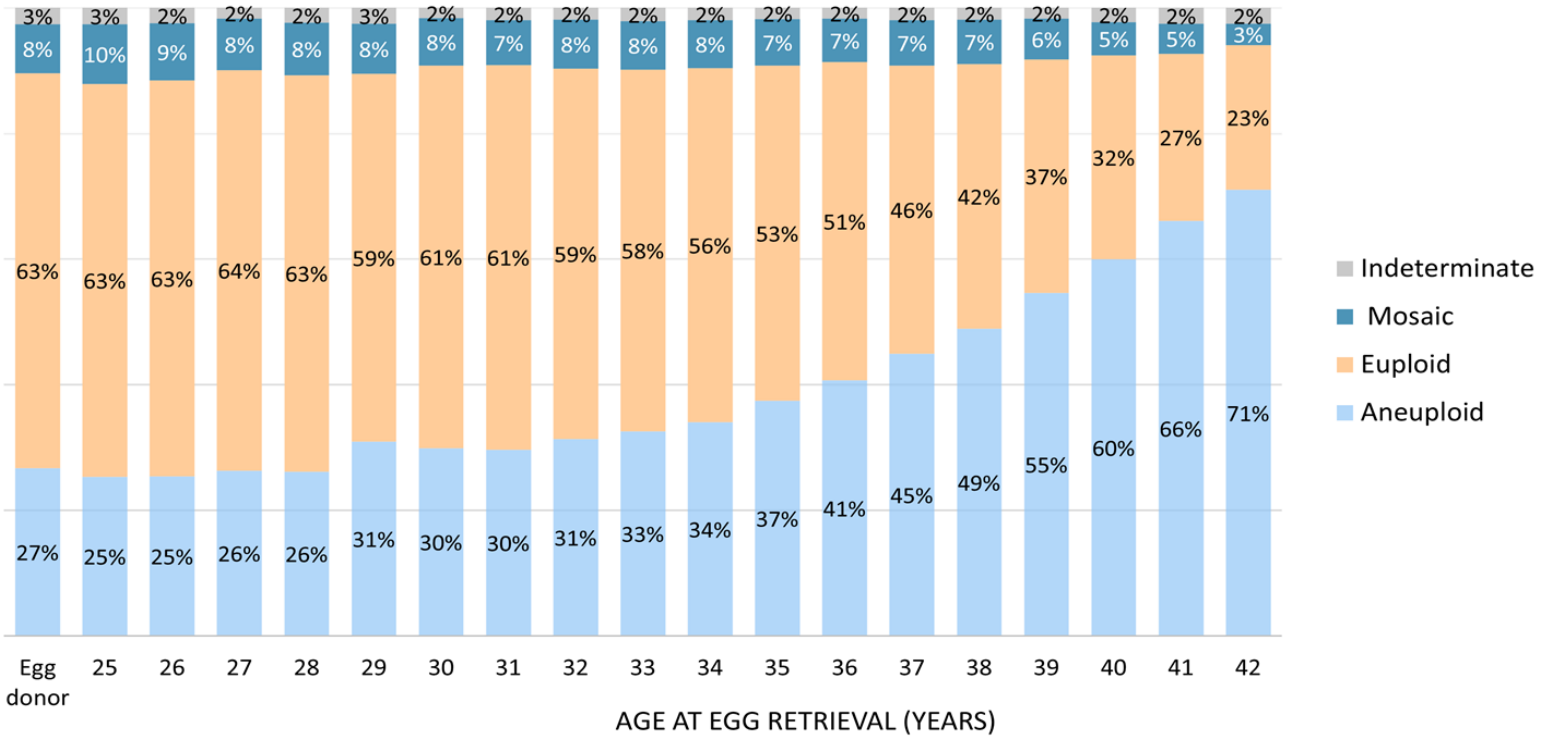
PGT-A BASICS (continued)

ANEUPLOIDY

Aneuploidy is typically sporadic, meaning it happens randomly. Aneuploidy is frequently detected in embryos, and it is typical to see some aneuploid and some euploid results on a PGT-A report.

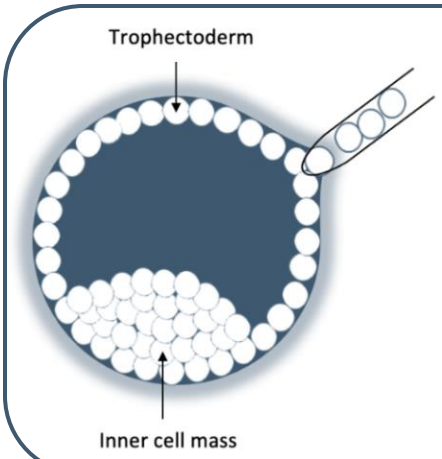
The chance for aneuploidy increases as age at the time of egg retrieval increases. The below graph presents internal Igenomix data on the chances for aneuploidy based on age at egg retrieval.

ANEUPLOIDY IN EMBRYOS BY AGE AT EGG RETRIEVAL



PGT-A BASICS (continued)

HOW IS ANEUPLOIDY ASSESSED ON PGT-A?

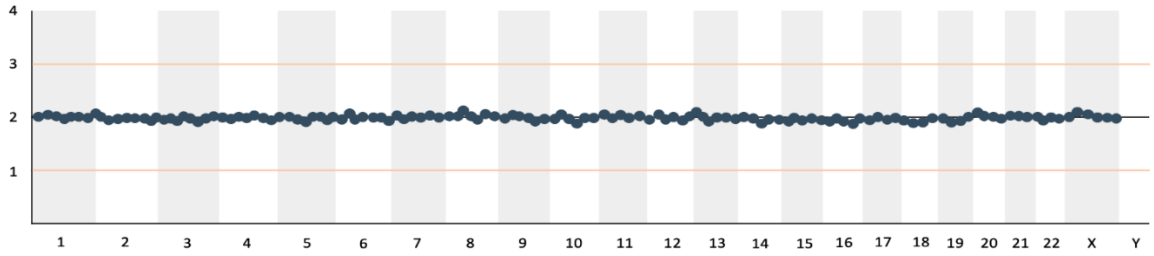


Testing a preimplantation embryo

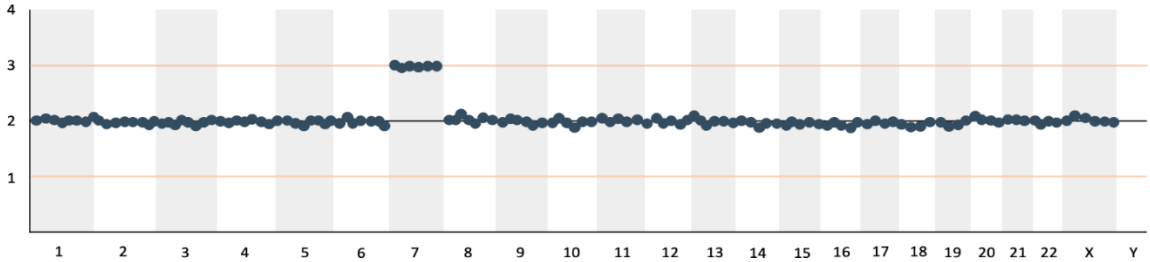
PGT-A involves testing a small sample of cells (a biopsy) from the embryo. Embryo biopsies are taken from the outer layer of the embryo. These cells are called the **trophectoderm** and they will eventually form the placenta. The part of the embryo that will eventually form the fetus, the **inner cell mass**, remains untouched.

Cells from the sample are analyzed together as a whole using next-generation sequencing (NGS) technology. Each cell cannot be analyzed individually.

A **euploid** result is reported when the number of sequencing reads, representing the amount of DNA, from each chromosome falls on the baseline, determined to be copy number 2.



Chromosome reads that deviate from copy number 2 are an indication of aneuploidy. An increase to copy number 3 (extra chromosome) or a decrease to copy number 1 (missing chromosome) is consistent with the presence of a chromosomal abnormality. These results are reported as **aneuploid**.



TYPES OF RESULTS

PGT FOR ANEUPLOIDY SCREENING (PGT-A)

RESULT: Euploid

WHAT IT MAY LOOK LIKE ON YOUR REPORT: Euploid

An embryo is reported as euploid (normal) if all 23 pairs of chromosomes are identified. Embryos reported as euploid are the best candidates for transfer. They are most likely to implant, most likely to develop to term, and most likely to result in a healthy livebirth. Euploid embryos will be bolded on the PGT-A report.

RESULT: Aneuploid

WHAT IT MAY LOOK LIKE ON YOUR REPORT:

Aneuploid: Trisomy 20 Aneuploid: Monosomy 14

Embryos are reported as aneuploid when NGS detects a copy number change signifying an extra or missing chromosome. Trisomy refers to an extra chromosome, and monosomy refers to a missing chromosome. For example, "Aneuploid: Trisomy 20" means that a gain of chromosome 20 was detected, indicating there are three copies of chromosome 20 instead of the typical two. "Aneuploid: Monosomy 5" means that a loss of chromosome 5 was detected, indicating there is only one copy of chromosome 5. Most types of aneuploidy are incompatible with life (nonviable). Aneuploid embryos will most likely miscarry early in the pregnancy or never implant. For more information on specific aneuploidies, please see the supplemental aneuploidy guide on the following pages.

RESULT: Complex Aneuploid

WHAT IT MAY LOOK LIKE ON YOUR REPORT:

Complex aneuploid: Trisomy 1 and 22, Monosomy 14
Complex aneuploid: Monosomy 5 and 19, Trisomy 6 and 12

Embryos are reported as complex aneuploid when there are 2-5 chromosome changes identified. The presence of multiple chromosome abnormalities would be expected to have a significant impact on development, and these embryos are expected to miscarry early during the pregnancy or never implant.

**Mosaic reporting is an option selected by the ordering provider. If your report includes mosaicism, the report will say "Analysis and Reporting: Full mosaic reporting" beneath the results. If your report does not include mosaicism, the report will say "Analysis and Reporting: Aneuploidy reported above 50% threshold." Questions about whether mosaic information would be included in your results can be discussed with your clinic. For additional information regarding the implications of a mosaic PGT-A result, please see our separate handout called "Mosaic Results Guide."*

RESULT: Mosaic*

WHAT IT MAY LOOK LIKE ON YOUR REPORT:

Low mosaic: Low mosaic trisomy 19

High mosaic: High mosaic partial trisomy 3q25.2q29 (44 Mb)

If some of the cells in an embryo have a chromosome change and others don't, then that embryo is mosaic. Embryos are reported as mosaic when NGS detects an *intermediate* copy number change, suggesting a mix of euploid and aneuploid cells in the biopsy. "Low mosaic" results have chromosomal copy number variation between 30-50%, and "high mosaic" embryos have chromosomal copy number variation between 50-70%. Studies continue to assess clinical outcomes following transfer of embryos reported as mosaic. Low mosaic embryos may have reproductive potential nearly equal to that of euploid embryos. High mosaic embryos may also have reproductive potential but higher risks for miscarriage compared to euploid embryos. The risk for mosaicism to persist throughout pregnancy is thought to be low.

RESULT: Complex Mosaic*

WHAT IT MAY LOOK LIKE ON YOUR REPORT:

Complex mosaic: Low mosaic trisomy 10 and 16

Complex mosaic: High mosaic trisomy 15 and 20, Low mosaic monosomy 2

Embryos are reported as complex mosaic when there are 2-5 chromosomes with intermediate copy number changes identified. Embryos reported as complex mosaic may include low mosaic chromosomes, high mosaic chromosomes, or a combination of both. Please see above "mosaic" explanation above for additional information regarding a mosaic result.

RESULT: Segmental Aneuploid

WHAT IT MAY LOOK LIKE ON YOUR REPORT:

Aneuploid: Partial monosomy 1q25.3q44 (65 Mb) or Aneuploid: Partial loss 1q25.3q44 (65 Mb)

Aneuploid: Partial trisomy 3p26.3p24.2 (24 Mb) or Aneuploid: Partial gain 3p26.3p24.2 (24 Mb)

A result of partial aneuploidy with a "p" or "q" following the chromosome number indicates segmental aneuploidy. Embryos are reported as segmental aneuploid when there is an extra or missing piece of a chromosome, rather than a whole chromosome imbalance. The "p" or "q" in the result refers to which part of the chromosome contains the extra or missing piece. The letter "p" refers to the top portion of the chromosome, also known as the short arm, and "q" refers to the bottom portion of the chromosome, also known as the long arm. The numbers following the "p" or "q" are the approximate "breakpoints," telling us where on the chromosome the imbalance occurs. The number in parentheses refers to the approximate size of the imbalance. One megabase (Mb) is one million base pairs, or one million letters of genetic code. Putting that all together, "Partial monosomy 1q25.3q44 (65 Mb)" means that there is a missing segment of chromosome 1. The missing segment is on the q-arm (bottom half) of the chromosome and is approximately 65 Mb in size.

Segmental aneuploidy can happen randomly and is usually not associated with a characterized genetic syndrome. Depending on the size and location of the imbalance, an embryo with uniform segmental aneuploidy would be expected to miscarry early, never implant, or lead to the birth of a child with a chromosome condition. Emerging evidence suggests that segmental aneuploidies are more likely to be mitotic in origin. This means that they could be mosaic and have reproductive potential, even if mosaicism is not detected in the biopsy sample. If the cells that develop into a fetus have a segmental aneuploidy, there could be a significant impact on development, so there are risks to consider.

RESULT: Chaotic

WHAT IT MAY LOOK LIKE ON YOUR REPORT: Aneuploid: Chaotic

Embryos are reported as chaotic when there are six or more chromosome changes identified. For chaotic aneuploid results, the individual chromosome changes identified are not listed on the report. If the abnormalities identified in the biopsy are present in the whole embryo, the embryo would be expected to miscarry early in the pregnancy or never implant. The predictive value of a chaotic aneuploid result may be reduced. This means it is possible that some, or potentially all, of the reported aneuploidies may not be truly present. Accordingly, re-biopsy and re-testing can be considered at the clinic's discretion. There is very limited clinical outcome data for embryos with chaotic results, so there are risks to consider.

RESULT: Indeterminate

WHAT IT MAY LOOK LIKE ON YOUR REPORT:

No DNA detected

Non-informative

An indeterminate result means that the laboratory was not able to obtain a reliable result for the embryo. An indeterminate result can happen if there is not enough DNA in the biopsy sample (No DNA detected) or the quality of DNA is not sufficient to get a result on that embryo (Non-informative). The embryo could be chromosomally normal or abnormal, and it would have the same, age-dependent chances for aneuploidy as an untested embryo. Re-biopsy and re-testing can be considered at the clinic's discretion and depending on embryo quality.

****

The following result type only applies for Smart PGT-A Plus reports. It should be noted that ploidy abnormalities cannot be detected with the standard Smart PGT-A platform.

RESULT: Abnormal Ploidy

WHAT IT MAY LOOK LIKE ON YOUR REPORT:

Triploid XXY

Haploid X

Typically, there are 2 sets of every chromosome (also known as diploid). A ploidy abnormality is when the number of chromosome sets is not 2. Embryos are reported as haploid when there is only one set of chromosomes detected (i.e. 23 total chromosomes instead of the typical 46). Embryos are reported as triploid when there are three sets of chromosomes detected (i.e. 69 total chromosomes instead of the typical 46). There may also be additional gains or losses of specific chromosomes indicated on the report. Embryos with triploidy or haploidy are likely to miscarry early during the pregnancy or never implant. Embryos with ploidy abnormalities can result in a molar pregnancy with risks to the individual carrying the pregnancy.

FREQUENTLY ASKED QUESTIONS

PGT FOR ANEUPLOIDY SCREENING (PGT-A)

GENERAL QUESTIONS

DOES A EUPLOID RESULT GUARANTEE A HEALTHY BABY?

Getting a euploid result is great news! Embryos reported as euploid have the best chances for resulting in a healthy, ongoing pregnancy. While embryos reported as euploid have the highest reproductive potential, PGT-A cannot guarantee the birth of a healthy child as there are many factors that impact implantation and fetal development.

WHAT DOES PGT-A TEST FOR?

PGT-A analyzes the chromosome content of embryos to assess if there are any chromosomes that are extra or missing. The goal of PGT-A is to identify embryos with the highest reproductive potential. PGT-A does not screen for every genetic disorder. PGT-A does not screen for single gene disorders like cystic fibrosis, sickle cell disease, or BRCA1/2, and it does not test for health conditions with both genetic and environmental contributions (multifactorial disorders) such as autism or autoimmune disorders. Additionally, PGT-A does not directly test for birth defects. There is a general 4-6% risk for birth defects for any baby born following IVF treatment.

WHAT TYPES OF CONDITIONS OR SYMPTOMS ARE EXPECTED FROM THESE ANEUPLOID RESULTS?

Please see the supplemental aneuploidy guide (p.18) for information regarding each specific aneuploidy. Only a handful of aneuploidies are associated with specific genetic conditions. This is because the majority of aneuploidies are not compatible with life, and therefore are not seen in ongoing pregnancies or live born babies. The goal of PGT-A is to test for aneuploidies that prevent pregnancy or the birth of a healthy baby, rather than just for those associated with syndromes.

ACCURACY

HOW ACCURATE IS PGT-A?

The accuracy of PGT-A is estimated to be 98%. This means there is an approximately 2% chance that PGT-A results may not reflect the chromosomal makeup of the embryo. This is largely due to a biological limitation of PGT-A, which is that testing is performed on only a small sample of cells and cannot assess the whole embryo.

SHOULD I DO PRENATAL GENETIC TESTING AFTER TRANSFERRING A PGT-A TESTED EMBRYO?

All genetic testing is optional; however, prenatal genetic testing can be considered after transfer of a PGT-A tested embryo in order to confirm the results. PGT-A does not replace prenatal testing. We recommend speaking with your obstetrician and/ or a prenatal genetic counselor about genetic testing options during pregnancy.

I'VE READ THAT ABNORMAL EMBRYOS CAN SELF-CORRECT AND RESULT IN HEALTHY BABIES. IS THIS TRUE?

This likely refers to emerging data regarding transfer of embryos reported as mosaic. One theory for this phenomenon is that the euploid (normal) cells in mosaic embryos preferentially replicate, while the aneuploid (abnormal) cells do not, which over time results in a chromosomally normal pregnancy. While individual aneuploid cells cannot be "corrected," a mosaic embryo may have the potential to result in a chromosomally normal baby. Since non-mosaic aneuploid embryos do not contain any euploid (normal) cells, they would not be expected to "self-correct" to a chromosomally normal pregnancy.

ANEUPLOIDY

ARE SIMILAR RESULTS EXPECTED IN FUTURE CYCLES?

Typically, results from one cycle do not have an impact on future cycles. The chance of aneuploidy is best predicted by age at the time of egg retrieval, and this chance increases gradually with age. The below chart shows the chances of getting at least one euploid embryo in a cycle, based on age at egg retrieval.

CHANCE FOR AT LEAST ONE EUPLOID EMBRYO	
Age at egg retrieval (years)	**Chance for 1+ euploid embryo / IVF cycle**
25-30	91%
31-34	89%
35-37	83%
38-40	68%
41-42	51%

I DID NOT HAVE ANY EUPLOID EMBRYOS IN THIS CYCLE. WHAT DOES THIS MEAN?

We recognize that these are not the results you were hoping for. While these results can be discouraging, they are typically due to random chance and are not cause for concern from a medical standpoint. The outcome of each IVF cycle is viewed independently and the proportion of aneuploid embryos in one cycle does not predict the proportion of aneuploid embryos expected in future cycles. In some rare cases, multiple embryos with the same chromosomal abnormality may be suggestive of a chromosome rearrangement in a parent, but in these cases, Igenomix will include a comment on the report with follow-up recommendations.

ANEUPLOIDY (CONTINUED)

IS THERE ANYTHING I CAN DO TO IMPROVE MY RESULTS FOR MY NEXT CYCLE?

Aneuploidy typically happens sporadically, meaning there is, unfortunately, nothing you can do in the future – and nothing you could have done for this cycle – to reduce your chances of creating an aneuploid embryo. Potential management changes to improve overall cycle outcomes can be discussed with your IVF providers.

DID THIS ANUEPLOIDY COME FROM THE EGG OR THE SPERM?

Aneuploidy typically occurs due to an error in the egg due to biological factors; however, aneuploidy can come from either the egg or the sperm. Parental origin of the aneuploidy cannot be determined by PGT-A testing with NGS.

IF I HAVE MORE THAN ONE EUPLOID EMBRYO, WHICH ONE IS "BETTER" FOR TRANSFER?

The decision of which embryo to transfer ultimately belongs to you and your physician. If there are multiple euploid embryos, morphology (embryo grading) may be used to help prioritize embryos for transfer. Embryo grading is determined by your fertility clinic and can be further discussed with your clinical team.

EMBRYO QC*

**Embryo QC is an additional assessment performed with Smart PGT-A Plus. It is not available with standard Smart PGT-A. Smart PGT-A Plus must be requested by the ordering provider prior to testing.*

What does Embryo QC mean?

Embryo QC, or quality control, is an additional assessment designed to increase the accuracy of PGT-A. Embryo QC assesses for potential contamination and provides reassurance that all embryos in the cohort are genetically related to each other (i.e. 'sibling relatedness' measure). A "Pass" on embryo QC assessment increases the confidence that the sample analyzed corresponds to the tested embryo. If contamination is detected, then the result might not represent the intended embryo, and rebiopsy may be considered, especially if the result is euploid. If a sibling mismatch is detected, a procedural error may have occurred, and rebiopsy may be considered. A partial pass ("Pass*") may be given if no contamination was detected but sibling relatedness could not be assessed due to a limitation, such as there being only a single embryo in the cohort.

GRADING/MORPHOLOGY

WHAT ABOUT THE GRADING/MORPHOLOGY OF MY EMBRYOS?

Embryo grading may be included on PGT-A reports at the request of the ordering provider. Embryo grading is determined by your IVF clinic. For all questions related to grading and morphology, we recommend reaching out to your IVF clinic.

MITOSCORE*

**MitoScore reporting is an option selected by the ordering provider. Questions about whether MitoScore information would be included in your results can be discussed with your clinic.*

WHAT IS MITOSCORE?

MitoScore is a test done on chromosomally normal (euploid) embryos only. If you have multiple euploid embryos with the same morphology, the MitoScore results can help to prioritize embryos for transfer. Embryos with normal chromosomes are good candidates for transfer regardless of the results of the MitoScore test. Embryos with lower MitoScore values will be ranked higher and embryos with higher MitoScore values will be ranked lower. Embryo grading should be prioritized over the MitoScore when deciding which embryos to transfer.

SUPPLEMENTAL ANEUPLOIDY GUIDE

PGT FOR ANEUPLOIDY SCREENING (PGT-A)

Please note that this aneuploidy guide only applies to embryos with single whole chromosome aneuploidies. This guide does not apply to embryos reported as mosaic, segmental, complex, or chaotic.

RESULT	WHAT THIS RESULT MEANS
Trisomy 1	This means that there is an extra copy of chromosome 1. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 1 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 1	This means that there is a missing copy of chromosome 1. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 1 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 2	This means that there is an extra copy of chromosome 2. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 2 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 2	This means that there is a missing copy of chromosome 2. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 2 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 3	This means that there is an extra copy of chromosome 3. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 3 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 3	This means that there is a missing copy of chromosome 3. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 3 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 4	This means that there is an extra copy of chromosome 4. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 4 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 4	This means that there is a missing copy of chromosome 4. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 4 were to be transferred, it is expected it would either not implant or result in early miscarriage.

**Viable aneuploidies associated with genetic syndromes are marked by an asterisk.*

RESULT	WHAT THIS RESULT MEANS
Trisomy 5	This means that there is an extra copy of chromosome 5. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 5 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 5	This means that there is a missing copy of chromosome 5. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 5 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 6	This means that there is an extra copy of chromosome 6. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 6 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 6	This means that there is a missing copy of chromosome 6. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 6 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 7	This means that there is an extra copy of chromosome 7. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 7 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 7	This means that there is a missing copy of chromosome 7. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 7 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 8	This means that there is an extra copy of chromosome 8. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 8 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 8	This means that there is a missing copy of chromosome 8. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 8 were to be transferred, it is expected it would either not implant or result in early miscarriage.

**Viable aneuploidies associated with genetic syndromes are marked by an asterisk.*

RESULT	WHAT THIS RESULT MEANS
Trisomy 9	This means that there is an extra copy of chromosome 9. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 9 were to be transferred, it is expected it would either not implant or result in early miscarriage. There are a few case reports of babies born with trisomy 9; however, this is exceptionally rare.
Monosomy 9	This means that there is a missing copy of chromosome 9. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 9 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 10	This means that there is an extra copy of chromosome 10. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 10 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 10	This means that there is a missing copy of chromosome 10. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 10 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 11	This means that there is an extra copy of chromosome 11. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 11 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 11	This means that there is a missing copy of chromosome 11. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 11 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 12	This means that there is an extra copy of chromosome 12. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 12 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 12	This means that there is a missing copy of chromosome 12. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 12 were to be transferred, it is expected it would either not implant or result in early miscarriage.

**Viable aneuploidies associated with genetic syndromes are marked by an asterisk.*

RESULT	WHAT THIS RESULT MEANS
Trisomy 13*	This means that the embryo has an extra copy of chromosome 13. This finding can be compatible with life, and causes a genetic condition called Patau syndrome. Although this condition can be seen in live-born babies, approximately 95% of embryos with trisomy 13 unfortunately miscarry during the pregnancy.
Monosomy 13	This means that there is a missing copy of chromosome 13. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 13 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 14	This means that there is an extra copy of chromosome 14. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 14 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 14	This means that there is a missing copy of chromosome 14. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 14 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 15	This means that there is an extra copy of chromosome 15. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 15 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 15	This means that there is a missing copy of chromosome 15. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 15 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 16	This means that there is an extra copy of chromosome 16. While trisomy 16 is not associated with a clinically described genetic syndrome, it is commonly seen in first trimester pregnancy losses. If an embryo with trisomy 16 were to be transferred, it could implant, but would be expected to result in early miscarriage.

**Viable aneuploidies associated with genetic syndromes are marked by an asterisk.*

RESULT	WHAT THIS RESULT MEANS
Monosomy 16	This means that there is a missing copy of chromosome 16. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 16 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 17	This means that there is an extra copy of chromosome 17. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 17 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 17	This means that there is a missing copy of chromosome 17. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 17 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 18*	This means that the embryo has an extra copy of chromosome 18. This finding can be compatible with life, and causes a genetic condition called Edwards syndrome. Although this condition can be seen in live-born babies, approximately 95% of embryos with trisomy 18 unfortunately miscarry during the pregnancy.
Monosomy 18	This means that there is a missing copy of chromosome 18. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 18 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 19	This means that there is an extra copy of chromosome 19. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 19 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy 19	This means that there is a missing copy of chromosome 19. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 19 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 20	This means that there is an extra copy of chromosome 20. This finding is not associated with a clinically described genetic syndrome. If an embryo with trisomy 20 were to be transferred, it is expected it would either not implant or result in early miscarriage.

**Viable aneuploidies associated with genetic syndromes are marked by an asterisk.*

RESULT	WHAT THIS RESULT MEANS
Monosomy 20	This means that there is a missing copy of chromosome 20. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 20 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 21*	This means that the embryo has an extra copy of chromosome 21. This finding can be compatible with life, and causes a genetic condition called Down syndrome. Although this condition can be seen in live-born babies, approximately 76% of embryos with trisomy 21 unfortunately miscarry during the pregnancy.
Monosomy 21	This means that there is a missing copy of chromosome 21. This finding is not associated with a clinically described genetic syndrome. If an embryo with monosomy 21 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Trisomy 22	This means that there is an extra copy of chromosome 22. While trisomy 22 is not associated with a clinically described genetic syndrome, it is commonly seen in first trimester pregnancy losses. If an embryo with trisomy 22 were to be transferred, it could implant, but would be expected to result in early miscarriage. There are a few case reports of babies born with trisomy 22; however, this is exceptionally rare.
Monosomy 22	This means that there is a missing copy of chromosome 22. This finding is not associated with a clinically described genetic syndrome. If an embryo monosomy 22 were to be transferred, it is expected it would either not implant or result in early miscarriage.
Monosomy X*	This means that the embryo only has one copy of the X chromosome. This finding can be compatible with life, and causes a genetic condition called Turner syndrome. Although this condition can be seen in live-born babies, approximately 99% of embryos with monosomy X unfortunately miscarry during the pregnancy.
Trisomy XXY*	This means that an extra copy of the X chromosome is present in a male embryo. This finding can be compatible with life and causes a genetic syndrome called Klinefelter syndrome; however, embryos with this finding may have a lower chance of implantation and/or higher chance of miscarriage compared to a euploid embryo.

**Viable aneuploidies associated with genetic syndromes are marked by an asterisk.*

RESULT	WHAT THIS RESULT MEANS
Trisomy XYY*	This means that an extra copy of the Y chromosome is present in a male embryo. This finding can be compatible with life and causes a genetic syndrome called XYY syndrome (sometimes referred to as Jacob's syndrome). XYY is not thought to be associated with a significantly increased chance of miscarriage.
Trisomy XXX*	This means that an extra copy of the X chromosome is present in a female embryo. This finding can be compatible with life and causes a genetic syndrome called Triple X syndrome. Triple X syndrome is not thought to be associated with a significantly increased chance of miscarriage.
Triploid XXX^ **Triploid XXY^** **Triploid XYY^**	This means that there is an additional set of chromosomes. Typically, there are 2 sets of every chromosome. Triploid embryos have 3 sets of every chromosome. While this finding is generally not compatible with life, triploidy can be associated with a partial molar pregnancy, with risks to the individual carrying the pregnancy. If a triploid embryo were to be transferred, it could implant, but would typically result in miscarriage.
Haploid X^	This means that there is a missing set of chromosomes. Typically, there are 2 sets of every chromosome. Haploid embryos have only 1 set of every chromosome. Embryos may be reported as haploid if there was a duplication of a single set of chromosomes, resulting in 2 sets of chromosomes, both coming from the same parent, instead of one from each parent. While this finding is generally not compatible with life, transfer of embryos reported as haploid could result in a complete molar pregnancy, with risks to the individual carrying the pregnancy.

**Viable aneuploidies associated with genetic syndromes are marked by an asterisk.*

^Ploidy abnormality results including "triploid" and "haploid" may be reported with Smart PGT-A Plus. Ploidy abnormalities are not able to be detected with standard Smart PGT-A.