Reproductive Potential of Abnormal Embryos with Whole Chromosome and Segmental Aneuploidies Diagnosed by NGS based PGT-A

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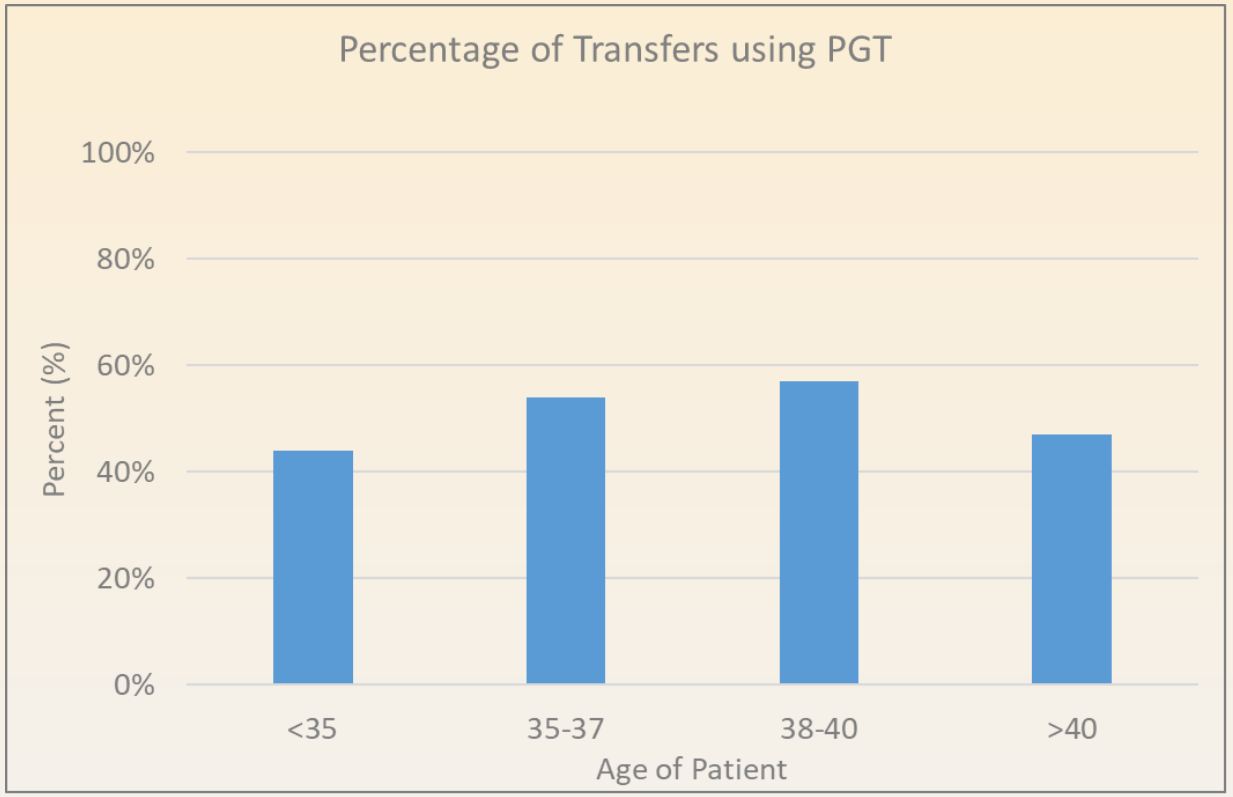
Financial Disclosure

I HAVE NO FINANCIAL OR COMMERCIAL CONFLICT OF INTEREST TO DISCLOSE

Preimplantation Genetic Testing (PGT-A)

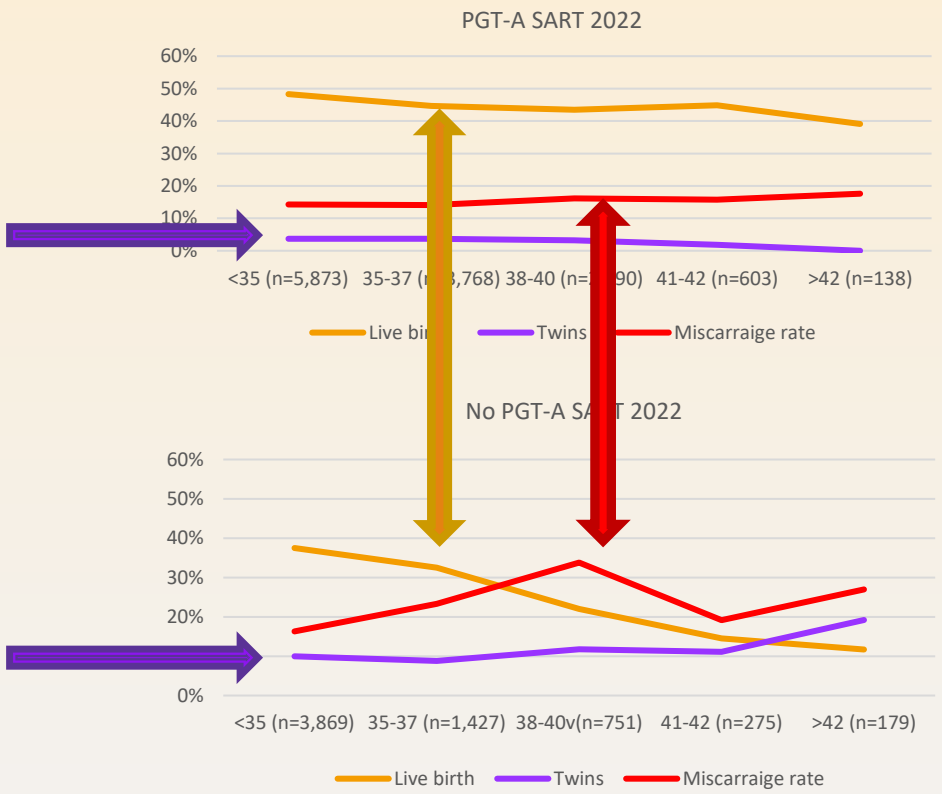
screening test of embryos for chromosomal abnormalities prior to ET

- ~2.5% of infants born in USA are conceived through ART
- > 50% of cycles in the USA use PGT-A to select euploid/mosaic embryos, to improve pregnancy rates per transfer, reducing miscarriages, and shortening time to LB



SART data based on 165,003 ART cycles , 2021

SART data 2022 PGT-A vs. non PGT-A



- Elective single embryo transfer can reduce multiple births and consequently can reduce maternal and neonatal adverse outcomes

FROZEN WITH PGT-A (BLASTOCYST)

Age of Woman at Retrieval	< 35	35 - 37	38 - 40	41 - 42	> 42
Number of Transfers	38159	26264	20282	6519	3026
Average Number of Embryos Transferred	1.0	1.0	1.0	1.0	1.0
% Cycles with Single Embryo Transfer	**96.1 %**	**96.9 %**	**97.3 %**	**97.4 %**	**96.0 %**
Live Birth Per Single Embryo Transfer	**52.4 %**	**51.5 %**	**50.0 %**	**48.7 %**	**44.4 %**

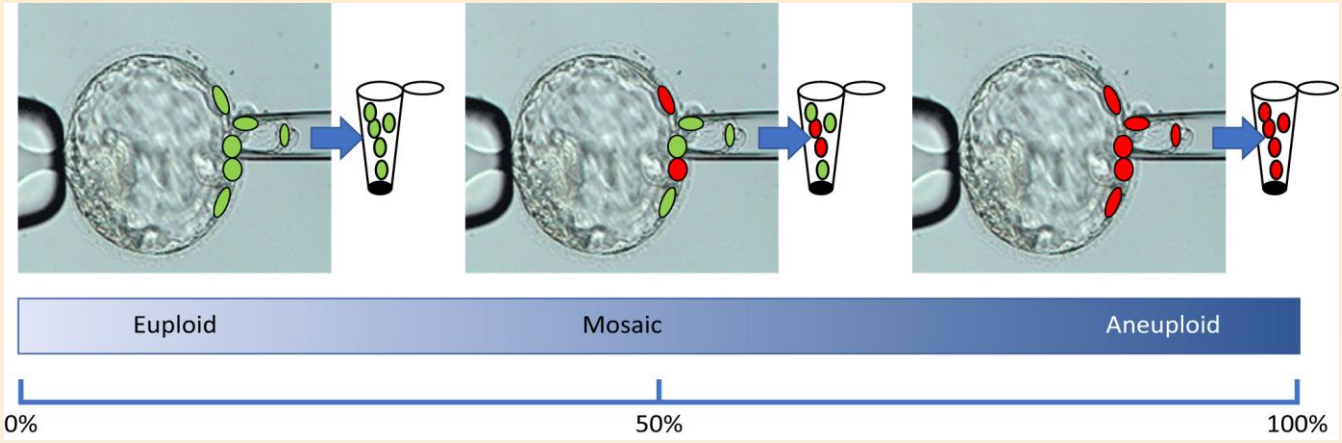
FROZEN NO PGT-A (BLASTOCYST)

Age of Woman at Retrieval	< 35	35 - 37	38 - 40	41 - 42	> 42
Number of Transfers	24920	10635	6537	2404	1859
Average Number of Embryos Transferred	1.1	1.2	1.3	1.5	1.7
% Cycles with Single Embryo Transfer	**85.5 %**	**80.5 %**	**70.9 %**	**60.7 %**	**58.0 %**
Live Birth Per Single Embryo Transfer	**39.8 %**	**32.2 %**	**23.0 %**	**16.9 %**	**12.7 %**

PGT-A

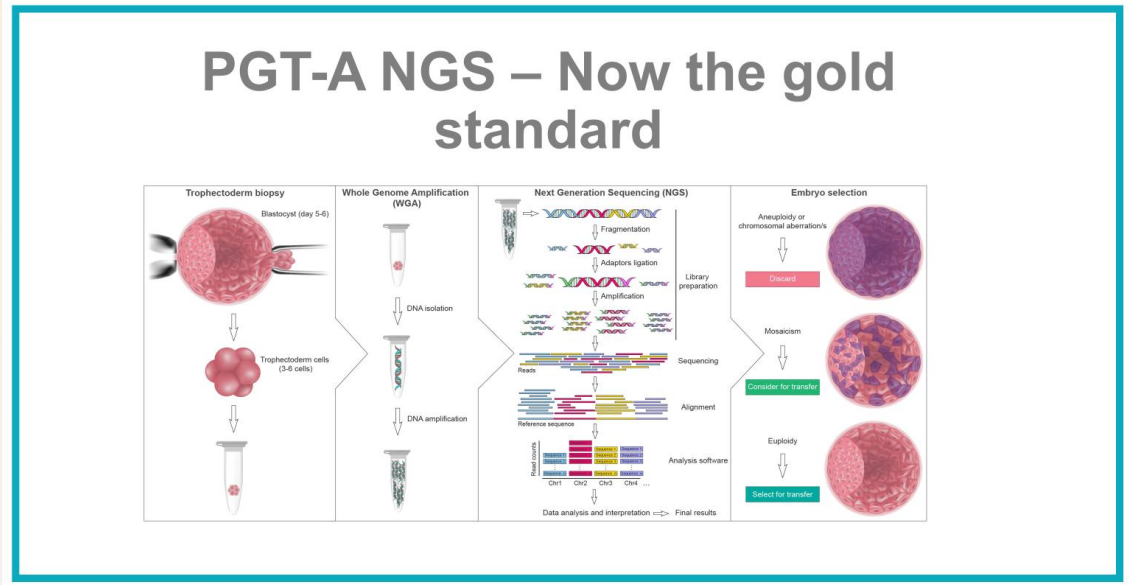
Embryo biopsy

- Day 5/6 (trophectoderm biopsy 3-6 cells)



Detection methods

- aCGH, (mosaicism detection rate >50%)
- **NGS,** (mosaicism detection rate >20%)
- Resolution to detect sub-chromosomal aberrations >10Mb

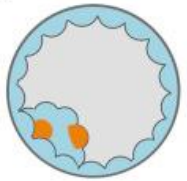


Limitations of PGT-A

Sampling bias – diagnostic accuracy (true representation of biopsied cells genome to the entire embryo and the inner cell mass **~96%**) Johnson DS et al, Hum Mol Reprod 2010, Victor A et al, Hum Reprod, 2018

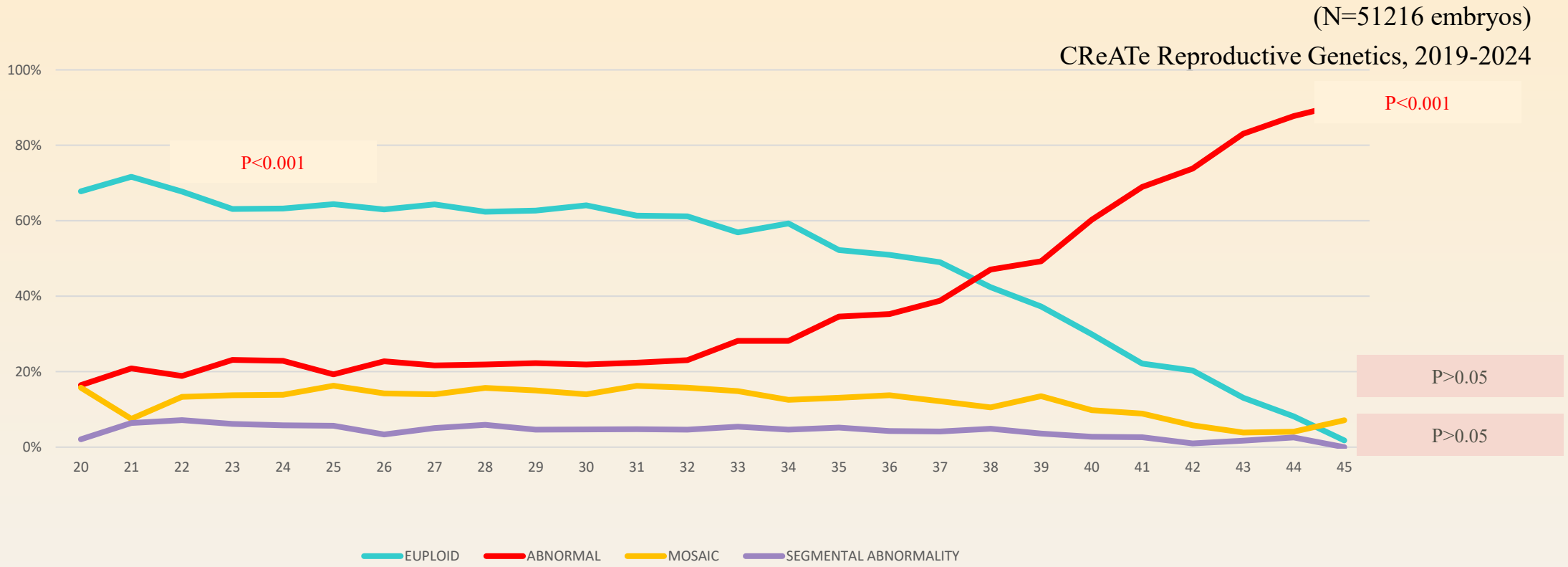
Diagnostic accuracy of PGT-A - testing platform dependent

- **98-100% Whole chromosomal aneuploidy** Kung A. et al. Reprod Biomed Online 2015, Kim J. et al. Fertil Steril 2022
  - Meiotic errors in oocytes are typically chromosome-specific, causing WCA, present in all cells of affected embryos, and generally, do not self-correct
  - Rare occurrence of uniparental disomy (UPD) in blastocyst-stage embryos (0.06%), reinforcing the rationale for using PGT-A to deselect embryos affected by these abnormalities to improve implantation and LB per transfer and reduce miscarriage rates
- **50-90% Mosaic embryo** Rodrigues V et al, Fertil Steril 2018, Maxwell SM et al, Fertil Steril 2016
- **~50% Segmental aneuploidies** Piccheta L. et al. F&S Science 2023

Mosaicism type	Possible TE biopsy	Diagnoses accuracy
Total Mosaic 	Euploid	Misdiagnosis
	Mosaic	Accurate
	Aneuploid	Misdiagnosis
ICM Mosaic 	Euploid	Misdiagnosis (Mosaicism never detectable)
TE Mosaic 	Euploid	Misdiagnosis
	Mosaic	Accurate
	Aneuploid	Misdiagnosis
ICM/TE Mosaic Type I 	Euploid	Misdiagnosis (Mosaicism never detectable)
ICM/TE Mosaic Type II 	Aneuploid	Misdiagnosis (Mosaicism never detectable)

Rodrigues V et al, 2018

Incidence of Aneuploidy by Oocyte Age Provider

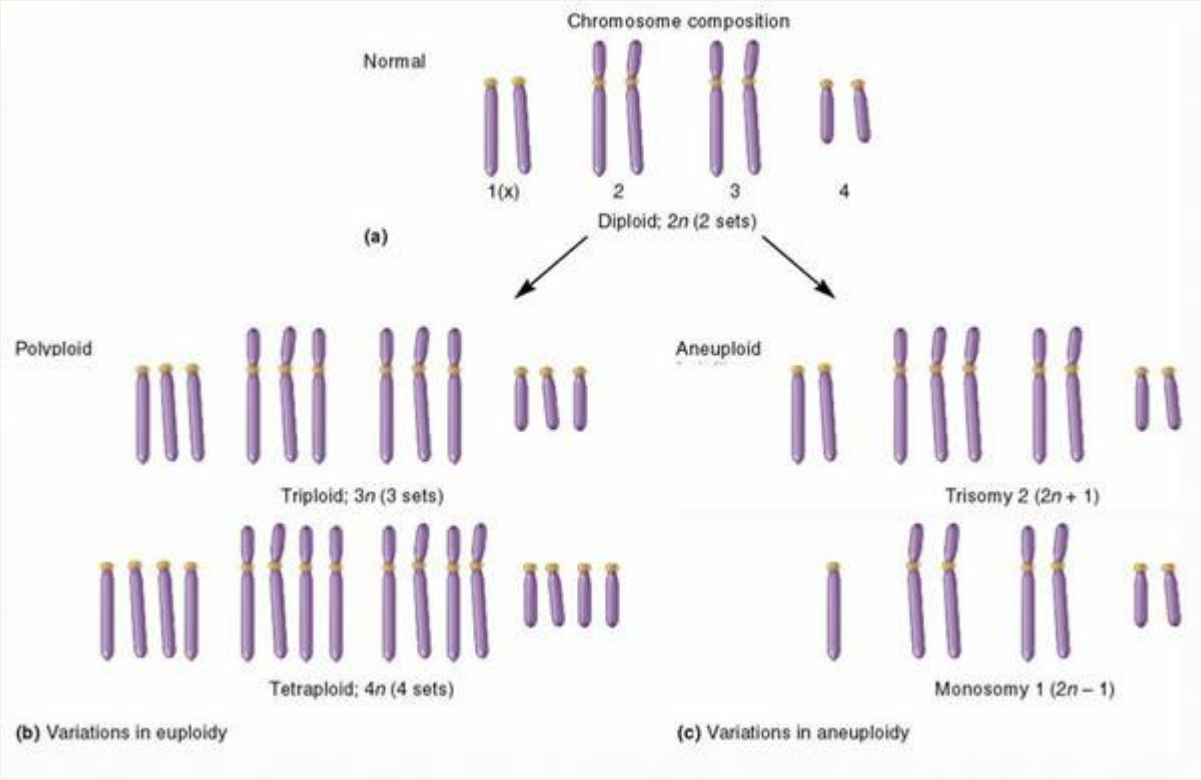


- **Whole Chromosome Aneuploidy** is strongly corelated to maternal age: incidence increases with advanced age
- **Mosaicism** is NOT related to maternal age: incidence declines with decline of euploid embryos with advanced age
- **Segmental aneuploidies** are NOT related to maternal age

Whole Chromosomal Aneuploidies (WCA)

represents abnormal number of chromosomes in the cell

- Primarily result from meiotic errors during chromosome segregation in gametogenesis,
- Occurrence is during oogenesis 90-99% and during spermatogenesis up to 10%
- Aneuploidy rates in oocytes have U-shaped are higher in women <20 years, decrease in adulthood, and rise significantly after age 35
  - Suggests age-related weakening of centromeric and systemic cohesion, causing premature chromatid segregation and reverse segregation with aging.
- Most WHA are incompatible with embryo development (non-implantation or early miscarriage)



Segmental Aneuploidy (SA)

are structural imbalances - gains or losses, involving a chromosomal segment

Meiotic or Mitotic in origin

Error in double strand break repair mechanisms (male germline more prone to SA because of high incidence of topoisomerase-mediated DSBs in spermatozoa DNA)

Sperm DNA damage predominantly leads to segmental imbalances, resulting in SA in embryos

No clinical guidelines on the use of SA embryos (yet!)

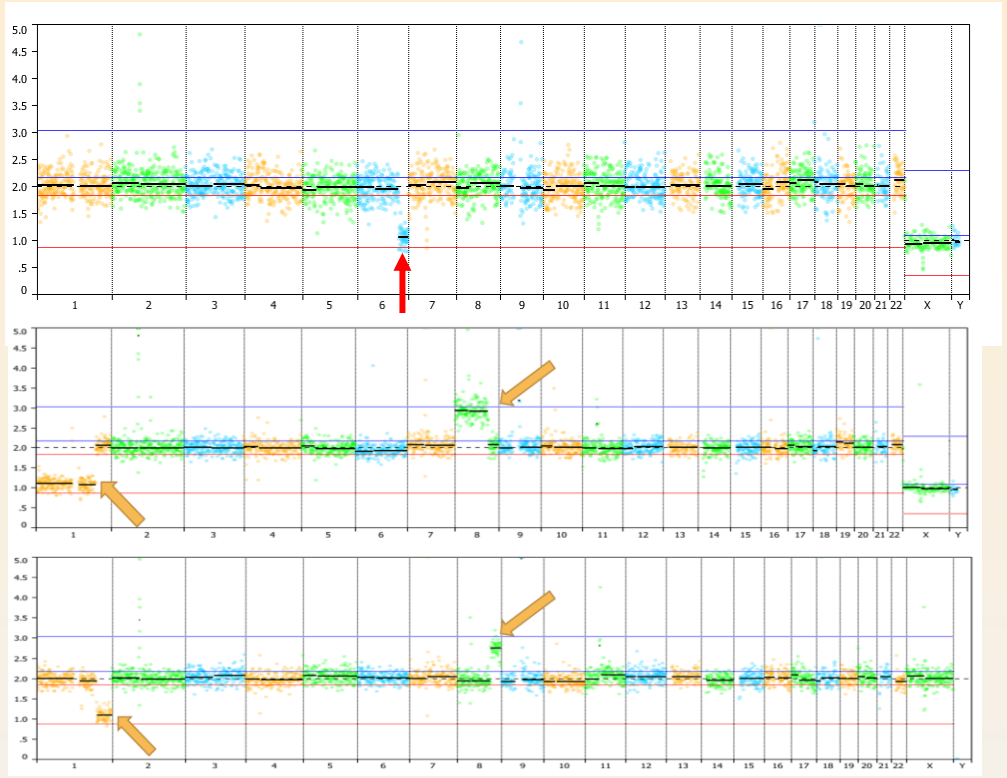


TABLE 1

Incidence of segmental aneuploidy in blastocyst-stage preimplantation embryos.

			Incidence of SA in blastocyst-stage embryos	
Reference	**Total no. of blastocysts**	**SA-positive blastocysts**	**Incidence of SA**	**Platform**
McCarty et al., 2022 (27)	89,226	2,766	3.10%	NGS
Babariya et al., 2017 (28)	1,327	207	15.60%	aCGH
Tiegs et al., 2021 (18)	2,110	186	8.82%	tNGS
Escribà et al., 2019 (26)	3,565	299	8.39%	NGS
Zore et al., 2019 (30)	377	20	5.31%	aCGH
Kubicek et al., 2019 (17)	967	54	5.58%	Karyomapping
Rechitsky et al., 2020 (35)	14,992	2,099	14.00%	NGS
Girardi et al., 2020 (24)	8,137	653	8.03%	NGS
Zuffardi et al., 2022 (31)	1,501	79	5.26%	NGS
Vialard et al., 2022 (32)	182,827	20,557	11,.24%	FAST-SeqS
Robberecht et al., 2021 (33)	1,708	97	5.68%	NGS
Zhou et al., 2018 (25)	2,095	206	9.83%	NGS
Xie et al., 2022 (21)	15,411	2,273	14.75%	NGS
Dviri et al., 2020 (34)	3,118	104	3.34%	NGS
Insua et al., 2018 (29)	3,628	314	[illegible]	NGS
Mean incidence of SA	-	-	8.51%	-

aCGH = array comparative genomic hybridization; NGS = next-generation sequencing; SA = segmental aneuploidy; tNGS = targeted next-g[illegible]ng.

Picchetta. Segmental aneuploidies in embryos. Fertil Steril Sci 2023.

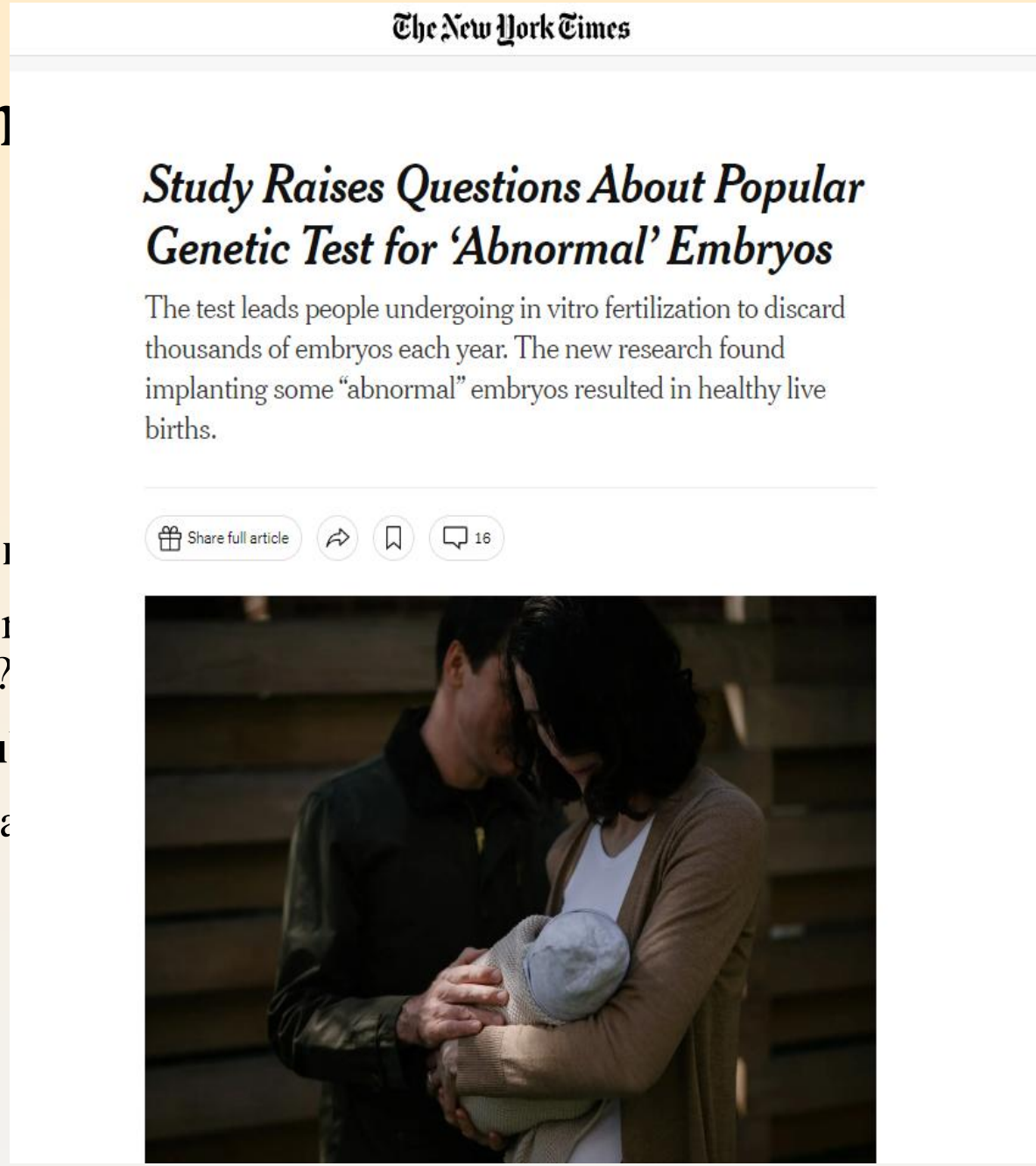
Picchetta L et al., 2023

What questions should we ask before the PGT-A testing

- What kind of **platfo**
- How the laboratory
  normal/transferable?
- Proper **review of pu**
- What are the expecta

/not-transferable or

tive LBR or LBR per transfer?



The New York Times

Study Raises Questions About Popular Genetic Test for 'Abnormal' Embryos

The test leads people undergoing in vitro fertilization to discard thousands of embryos each year. The new research found implanting some "abnormal" embryos resulted in healthy live births.

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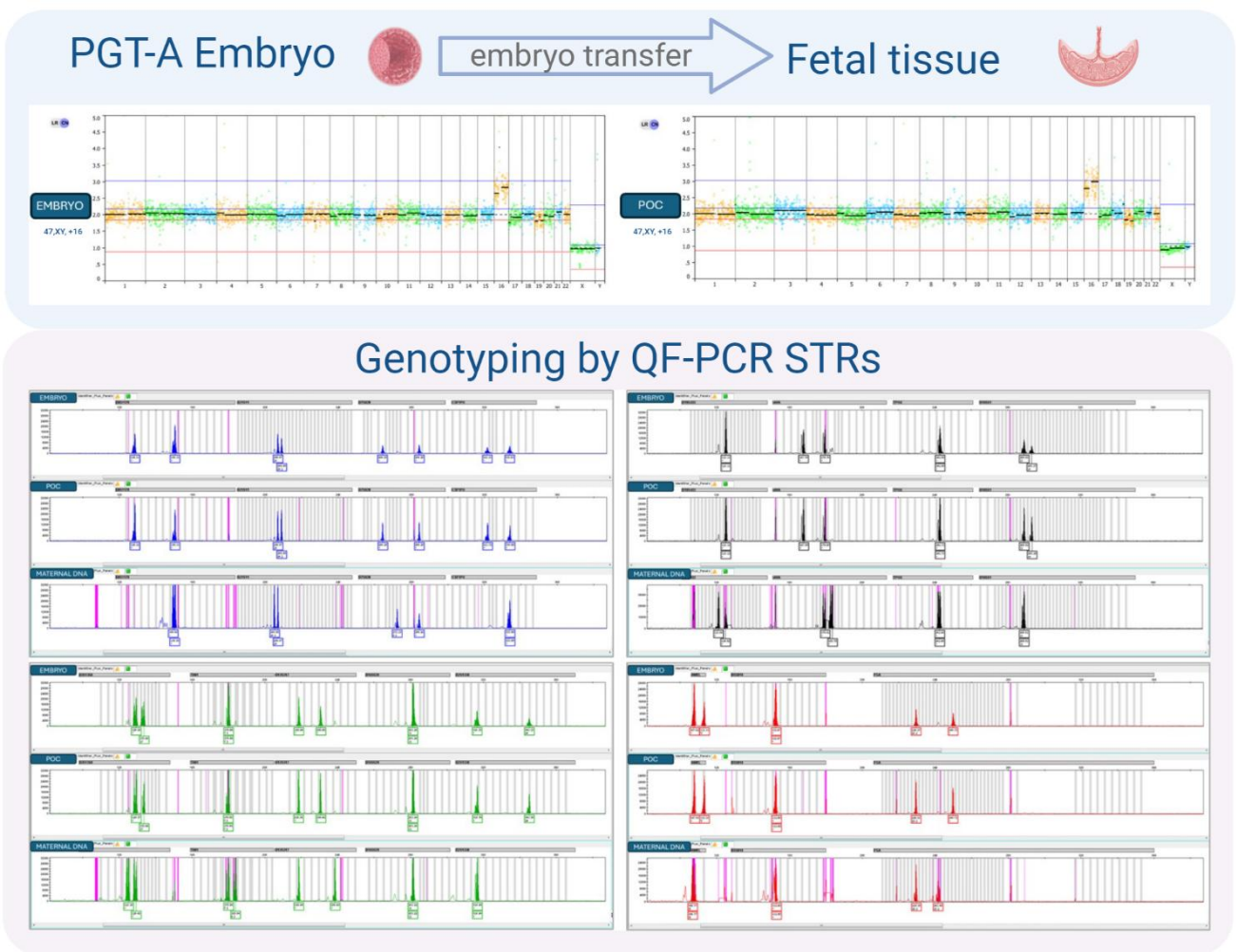
Identification of fetal identity and maternal cell contamination in products of conception



- Copy number variation plots from PGT-A NGS testing of DNA from trophectoderm biopsy and DNA from products of conception.
- Results show identical profile of embryo with trisomy 16 (47, XY, +16) and product of conception with trisomy 16 (47, XY, +16) after ET.
- Maternal cell contamination and confirmation of identity of transferred embryo was done by genotyping using highly informative STRs (AmpFLSTR Identifiler Plus PCR Amplification Kit (Life Technologies, Thermo Fisher Scientific).
- Results show identical STR profiles of embryo and POC and confirmation of no maternal cell contamination in the tested sample.

Important:

Genotyping after ET, confirms the same genome between PGT-A biopsy results and the Product of Conception, Fetus, or Baby, and it can exclude IVF lab, technical, PGT error, or natural conception.

A multicenter, prospective, blinded, nonselection study evaluating the predictive value of an aneuploid diagnosis using a targeted next-generation sequencing–based preimplantation genetic testing aneuploidy assay and impact of biopsy

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Brent Hanson, M.D.,[a,b] Emily Osman, M.D.,[a,b] Thomas J. Kim, M.D.,[d] George Patounakis, M.D., P
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ORIGINAL ARTICLE

PRENATAL DIAGNOSIS WILEY

IVF embryo choices and pregnancy outcomes

Li Wang[1,2] | Xiaohong Wang[3] | Yun Liu[4] | Xianghong Ou[5] | Min Li[6] |
Lei Chen[6] | Xiaoguang Shao[7] | Song Quan[8] | Jinliang Duan[9] | Wei He[10] |
Huan Shen[11] | Ling Sun[12] | Yuexin Yu[13] | David S. Cram[1] | Donald Leigh[1] |

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nature cell biology

AMENDMENTS

https://doi.org/10.1038/s41556-021-00775-x

Check for updates

Author Correction: Depletion of aneuploid cells in human embryos and gastruloids

Min Yang, Tiago Rito, Jakob Metzger, Jeffrey Naftaly, Rohan Soman, Jianjun Hu, David F. Albertini, David H. Barad, Ali H. Brivanlou and Norbert Gleicher

Correction to *Nature Cell Biology* https://doi.org/10.1038/s41556-021-00660-7, published 9 April 2021.

In the version of this Letter originally published online, Supplementary Table 1 included 9 of the 32 patients in this study who received chromosomally 'abnormal' embryos. The originally published table included only those 9 patients who achieved pregnancy. We here provide an updated and corrected version of the table including all 32 patients. In addition, during the manuscript preparation process, a transcription error regarding patient 7 occurred, listing one of the transferred embryos as 47,XX,+21 (Down syndrome), which we would not recommend for transfer and did not transfer. The actual transferred embryo was a mosaic trisomy-9 embryo with karyotype 47,XX,+9 [mos], which has now been corrected in the revised Supplementary Table 1. We furthermore received clinically relevant updates on patients 1 and 5. Patient 5 upon amniocentesis was initially reported as having a normal 46,XY pregnancy. Only after the foetus was diagnosed in utero by ultrasound with a coarctation of the aorta were further chromosomal studies done, which identified the deletion originally reported in a transferred embryo. The chromosomal status of the newborn was therefore corrected in the revised Supplementary Table 1, with the newborn deemed healthy after neonatal correction of the coarctation. Finally, based on oral reports from the patient, the information for patient 1 was amended to indicate embryos with karyotypes of 45,XY,−22 and 47,XY,+12. When a subsequent written report was received, the embryos were listed as 45,XX,−22 and 47,XY,+12 [mos], now reflected in a further correction made in the revised Supplementary Table 1. None of these changes affect the conclusions reached in the manuscript.

The original Letter has been corrected in the online version of the paper.

Published online: 21 September 2021
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LETTERS

https://doi.org/10.1038/s41556-021-00660-7

Check for updates

Depletion of aneuploid cells in human embryos and gastruloids

Min Yang[1,2,5], Tiago Rito[1,5], Jakob Metzger[1], Jeffrey Naftaly[1], Rohan Soman[1], Jianjun Hu[2], David F. Albertini[2], David H. Barad[2,3], Ali H. Brivanlou[1] and Norbert Gleicher[1,2,3,4]

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Advance Access Publication on April 12, 2022 https://doi.org/10.1093/humrep/deac063

human reproduction

ORIGINAL ARTICLE *Infertility*

IVF outcomes of embryos with abnormal PGT-A biopsy previously refused transfer: a prospective cohort study

D.H. Barad [1,2,*], D.F. Albertini[1], E. Molinari[1], and N. Gleicher[1,2,3,4]

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Submitted on October 27, 2021; resubmitted on February 26, 2022; editorial decision on March 21, 2022

What is the real reproductive potential of Abnormal Embryos?

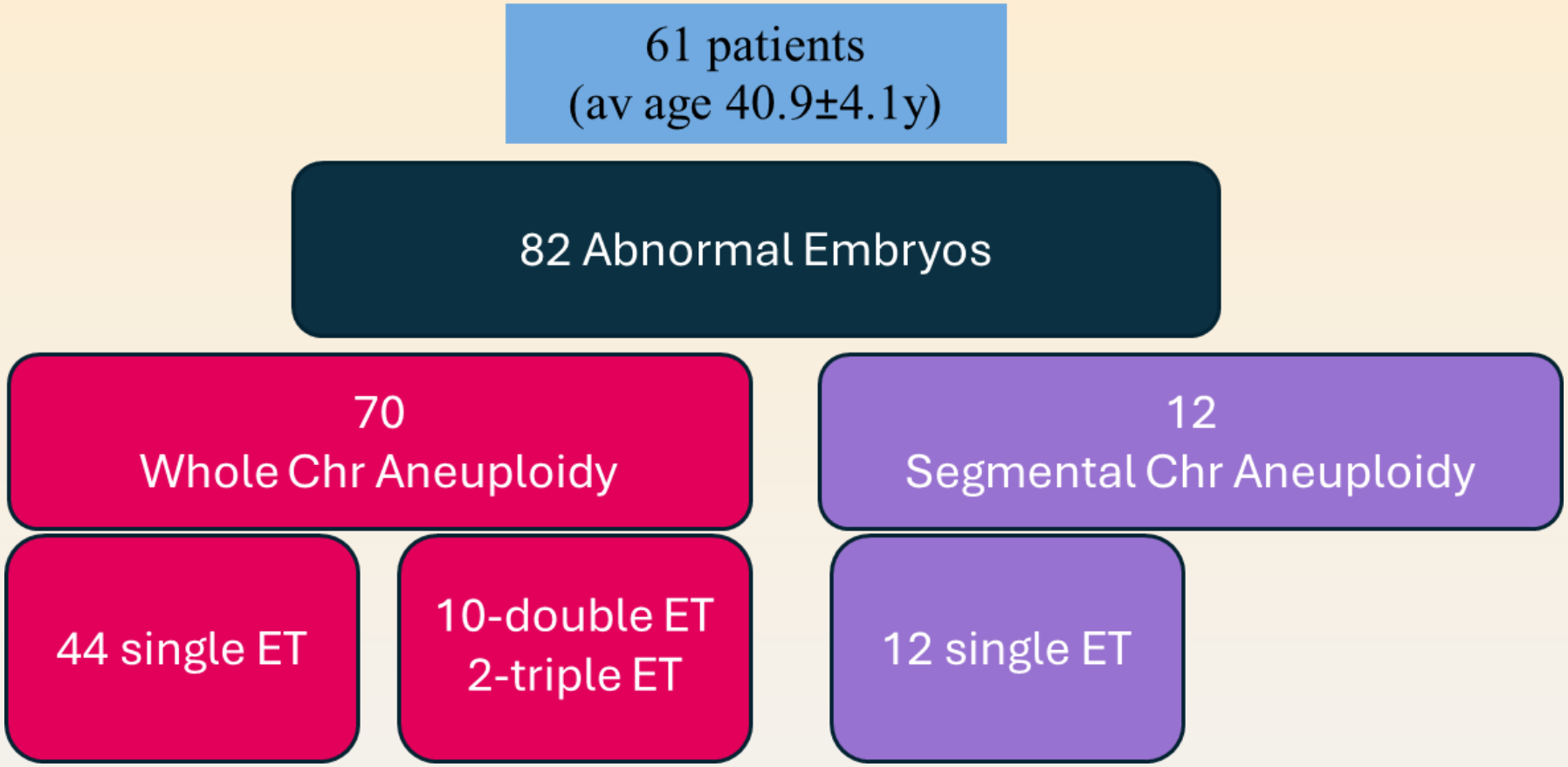
Aneuploid Embryo Transfer - Study design

- A retrospective, blinded, single center, cohort study (2016-2024)
- **99% of embryo transfers** were done ***without knowing the PGT-A results***
- TE-biopsy was performed before vitrification
- Patients' reasons for aneuploid transfer:
  - **Patient did fresh transfer** (PGT-A results were available after the pregnancy test result)
  - **Patient declined PGT-A testing at the time of transfer** (biobanked TE-biopsy sample was tested retrospectively)
  - ***Patient had an Abnormal Embryo transfer** (only one patient)

PGT-A test results were retrospectively collected from the Genetics database after assessing clinical outcomes

- **Primary outcomes: implantation rate (IR), miscarriage (MR) and live birth rate (LBR).**
- Baseline patient characteristics and outcome measures were analyzed using descriptive statistics.
- **Clinical outcomes were compared with PGT-A results to determine the predictive value of aneuploidy test result**

Patients and embryo transfer information



ET=embryo transfer.

Clinical outcomes after transfer of aneuploid embryos diagnosed by NGS based PGT-A

Transfer and pregnancy outcome	Whole Chromosomal Aneuploidies (n=70)	Segmental Aneuploidies (n=12)	P (<0.05 significant)
Implantation	14 (20%)	4 (33%)	NS
Biochemical pregnancy	5 (7%)	0	NS
Miscarriage*	9 (13%)	1 (8%)	NS
Ongoing pregnancy (normal prenatal testing) $	0	3 (25%)	0.004

Chromosomal Aberrations (monosomy –M/ trisomy –T)

WCA Biochemical pregnancy:

60% (3/5) had **monosomy** (M21-1, M15-1 and M11+M21)

40% (2/5) (had 2 aberrant chromosomes M/T (M11+T22 and T2+T16).

All WCA embryos that resulted in miscarriage had **trisomy**:

89% (8/9) single T (T22-1, T21-1, T18-1, T16-1, T15-1, T12-1, T-10-1?, T9-1 and

11% (1/9) multiple T chromosomes - T2+T14+T16+T18

NS=not significant

*Average gestational age is 7weeks 3days ±3days

$Healthy babies delivered at term (n=2), phenotypically normal. Cytogenetic testing was electively not performed. One ongoing pregnancy (second trimester at the time of writing) with normal first trimester screening results.

Whole Chromosome Aneuploid Embryo Transfers resulting in Miscarriage

Patient Number	Age	Biopsy Day	Grade	NGS PGT-A result	Reason for aneuploid transfer	FET DATE	Clinical Pregnancy outcome	Gestational age of miscarriage (w=weeks; d=days)	Follow up genetic testing
2	40	5	2BB	XY: +18	FRESH TRANSFER	23-Jan-18	Termination	12w4d	XY: +18
7	39	6	2BB	XX: +2; +14; +16; +18; (+9, 65%)	DECLINED TESTING	27-Oct-19	Miscarriage	5w	no
9	44	6	1BB	XY: +21	DECLINED TESTING	28-Sep-19	Miscarriage	9w	XY: +21
15	41	6	1BB	XY: +12	DECLINED TESTING	5-Oct-16	Miscarriage	6w	XY: +12
17	46	6	1BA	XY: +10	DECLINED TESTING	18-Apr-24	Miscarriage	10w	no
24	37	6	1BB	XX: +15	DECLINED TESTING	1-Apr-24	Miscarriage	5w3d	no
26	40	5	1BB	XY: +16	DECLINED TESTING	16-Dec-20	Miscarriage	7w6d	XY: +16
29	30	6	1BA	XY: +9	DECLINED TESTING	26-Jan-21	Miscarriage	6w1d	XY: +9
38	38	5	1CB	XY: +22	DECLINED TESTING	28-Jul-19	Miscarriage	9w4d	no

Segmental Aneuploid Embryo Transfers Outcomes

Patient Number	Age	Biopsy Day	Grade	NGS PGT-A result	Reason for aneuploid transfer	FET DATE	Clinical Pregnancy outcome	Gestational age of miscarriage (w=weeks; d=days)	Follow up genetic testing
1	48	6	1BB	XX: -3q22.1-q29|65.3Mb	DECLINED TESTING	20-Jan-18	Live birth		no
4	43	7	2DD	XY: -2p23.2-p11.2|29.9Mb; -11q14.3-q22.3|15.1Mb	DECLINED TESTING	21-Apr-23	N		
6	33	6	1BC	XX: -2q37.1-q37.3|11.3Mb	DECLINED TESTING	21-Feb-23	N		
26	40	6	1CB	XX: -7q32.1-q36.3|30.36Mb	DECLINED TESTING	24-May-21	N		
34	32	6	1BB	XY: -18q21.1-q23|33.1Mb	DECLINED TESTING	19-Mar-24	N		
43	40.3	6	1CC	XY: -1p36.33-p36.22|12.1Mb; +1p36.22-p21.1|94.6Mb	DECLINED TESTING	19-Oct-23	Live birth		no
44	38.1	6	1CC	XX: -1q|	DECLINED TESTING	17-Feb-23	Miscarriage	7w6d	no
45	39.7	7	3DD	XY: +Xp22.33|18Mb	DECLINED TESTING	8-Dec-22	N		
46	33.3	6	1BC	XX: -2q37.1-q37.3|11.3Mb	DECLINED TESTING	21-Feb-23	N		
48	33.5	6	2AB	XY: -10q23.1-q26.3|52Mb	DECLINED TESTING	31-Jul-24	N		
49	37.2	7	1CD	XX: -2p|	DECLINED TESTING	10-Sep-24	Ongoing pregnancy		
52	41.8	6	1BC	XX: -12p13.3-p13.2|12Mb; (-12p13.2-q24.3, 121.8Mb, 65%); (-13, 65%)	DECLINED TESTING	27-Sep-24	N		

Summarized outcomes from previous studies and the current study after transferring NGS PGT-A diagnosed aneuploid embryos

Study	PGT-A method	Whole Chromosome Aneuploidy ET	Livebirth rate after Aneuploid ET (%)	Miscarriage rate Aneuploid ET (%)	Segmental Aneuploidy ET	Livebirth rate after Segmental Aneuploid ET (%)
Tiegs et al., 2021	NGS	102	0/102 (0%)	100% (24/24)	0[@]	0
Wang et al., 2021	NGS	44	2[#]/44 (4.5%)	75% (6/8[#])	9	22% (2/9)
Barad et al, 2022 (Yang et al,. 2021)* [$]	NGS[$]	84[$]	0/84 (0%)[$]	100% (8/8^)	3	33% (1/3)
Besser et al, 2024	NGS	-	-	-	21	24% (5/21)
Madjunkov M et al, 2024	NGS	70	0/70	100% (9/9)	12	25% (3/12)
Total	NGS	300	2/300 (0.66%)[#]	95.9% (47/49)	45	24.4% (11/45)

[@] Tiegs et al. had blinded transfer of 39 embryos with "SCAs", however did not differentiate between mosaic SCAs and non-mosaic SCAs, hence excluded from this summary table.

*Barad et al. included the same embryo transfers reported by Yang et al

[$]The diagnostic platform for the embryo with PGT-A result 46, XX: +14 -18 born as 46, XX from Barad et al, 2022 was changed to microarray in a follow up publication by the same group Gleicher et al., 2023

^The number indicates patients (n=8) that had a total of sixteen abnormal embryos transferred, all tested by NGS. Among these patients, four had confirmed aneuploidy in fetal tissue, while the rest did not undergo genetic testing.

[#]Explanation of non-concordant transfer outcome(s) includes true mosaics (trophectoderm/ICM), sample mix up, errors in analysis or natural pregnancy. Additional genetic testing (genotyping) and comparison between biopsied material and liveborn kids should give an answer for these two cases, however these have not been performed.

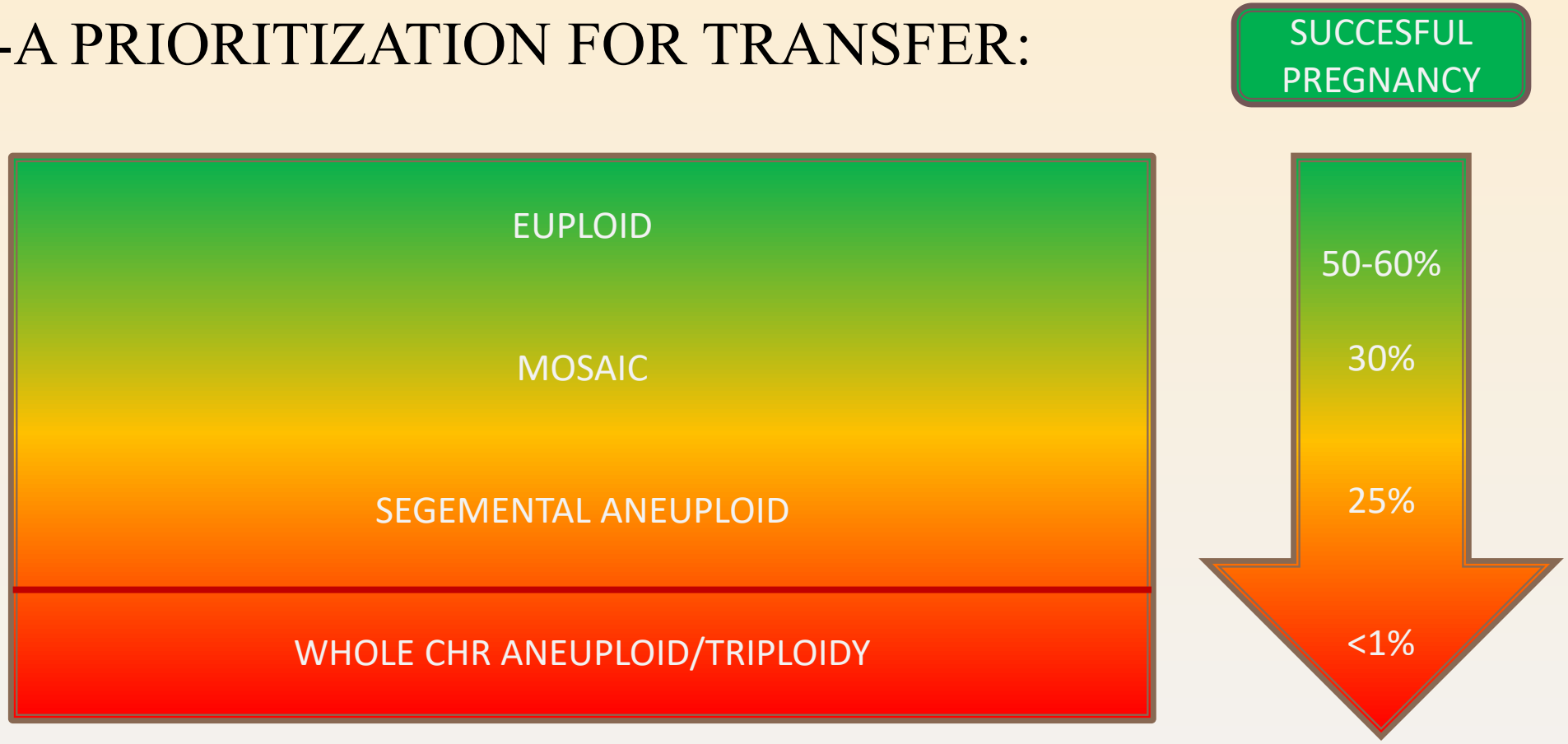
ET=embryo transfer

FINAL CONCLUSIONS

- ***Embryos exhibiting whole chr. aneuploidy have limited potential for development and the published data suggest that they not result in live birth***
- ***Using NGS based PGT-A to avoid transferring abnormal embryos with WCA can improve ART outcomes by reducing miscarriages, pregnancy terminations due to fetal aneuploidies, and failed transfers.***
- ***Transfer of segmental chrom. aneuploidy led to live births in ~25% of cases, consistent with previous reports.***
- ***Embryos with segmental aneuploidy should be considered a separate category of embryos with considerable reproductive potential.***
- ***All patients should receive close monitoring and genetic counseling.***

Are we ready to change guidelines on SA?

- PGT-A PRIORITIZATION FOR TRANSFER:



Acknowledgements

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Thank you!

Patient Number	Age	Biopsy Day	Grade	NGS PGT-A result	Reason for aneuploid transfer	FET DATE	Clinical Pregnancy outcome
Whole Chromosome Aneuploid Embryo Transfers resulting in No implantation (N)/Biochemical pregnancy							
3	43	6	1DC	XY: +2; +22	DECLINED TESTING	14-Mar-21	N
5	43	6	1CC	XY: +4; +16	DECLINED TESTING	27-May-19	N
5	43	5	2BB	XX: -13; -22	DECLINED TESTING	27-May-19	N
5-1	43	6	1DD	XY: -2	DECLINED TESTING	27-Jul-19	N
5-1	43	6	1CC	XX: -13; -22	DECLINED TESTING	27-Jul-19	N
5-1	43	5	1BB	XY: +13; -16	DECLINED TESTING	27-Jul-19	N
8	42	6	1BB	XY: +2; +16	DECLINED TESTING	23-Oct-23	Biochemical
10	36	6	2BC	XX: -15; -22	DECLINED TESTING	12-Aug-22	N
11	46	6	1BB	XX: -8; +16	DECLINED TESTING	9-Feb-24	N
11-1	46	6	2BB	XY: -16; (+7, 40%)	DECLINED TESTING	11-Apr-24	N
11-1	46	6	1BB	XY: -16; -22	DECLINED TESTING	11-Apr-24	N
12	33	6	1CB	XX: -4	DECLINED TESTING	30-Oct-20	N
13	43	7	2DC	XX: -18; +19	DECLINED TESTING	19-Feb-23	N
13	43	7	1CC	XY: +3p14.1-q29|128.6Mb; -17p|; +17q|; -21	DECLINED TESTING	19-Feb-23	N
14	40	6	1BC	XX: -7; -11	DECLINED TESTING	9-Jan-24	N
16	39	6	1BB	XX: -21	DECLINED TESTING	4-Nov-22	N
18	38	7	3CC	XY: -19; (+13, 30%)	DECLINED TESTING	26-Jul-21	N
19	42	5	1DD	XY: +6; +9; +18; (+19, 50%)	REPORT WASN'T SENT	25-Jan-21	N
20	32	6	1BA	XY: -22	DECLINED TESTING	23-Dec-19	N
21	42	6	1BB	XX: -21	DECLINED TESTING	13-Apr-22	N
21	42	7	1DD	XX: +15 (4 COPIES)	DECLINED TESTING	13-Apr-22	N
21-1	42	6	1CC	XY: -19;+22	DECLINED TESTING	20-Jun-22	N
21-1	42	6	1DB	XX: -8q12.3-q24.3|81.75Mb; -20; +22	DECLINED TESTING	20-Jun-22	N
22	43	6	2CC	XX: -16; -22	DECLINED TESTING	26-Apr-24	N
23	43	6	1BC	XX: -2	DECLINED TESTING	18-Feb-24	N
23	43	6	2BB	XY: -8; -21	DECLINED TESTING	18-Feb-24	N
25	43	6	2CC	XY: -4; (-3q, 50%); (+21, 50%); (-19, 40%)	DECLINED TESTING	1-Mar-24	N
26	40	6	1BB	XY: -20	DECLINED TESTING	19-Apr-22	N
27	43	7	1DD	XY: +5;-15;-21	DECLINED TESTING	16-Nov-21	N
28	39	7	3DD	XX: -8; +9; -21	ONLY SEX DISCLOSURE	19-Jul-21	N
28	39	6	2CC	XY: -16	ONLY SEX DISCLOSURE	20-Aug-21	N
30	35	6	2DC	XY: -5; +9; +12; +13; +16; +17; -21; -22	DECLINED TESTING	13-Feb-24	N
31	40	5	2BB	XX: +16	DECLINED TESTING	18-Aug-20	N
32	45	5	1BB	XX: +20	DECLINED TESTING	30-Nov-20	N
33	44	5	1BB	XY: +22; (-14, 30%)	DECLINED TESTING	27-Dec-20	N
33	44	5	1CB	XX: +15	DECLINED TESTING	27-Dec-20	N
33	44	5	1BB	XX: +12; (+1, 55%)	DECLINED TESTING	27-Dec-20	N
33-2	46	6	1BB	XX: -11; -21	DECLINED TESTING	18-Apr-21	Biochemical
33-2	46	6	2DD	XY: -11; +22	DECLINED TESTING	18-Apr-21	Biochemical
33-3	43	6	2CC	XX: +15	DECLINED TESTING	20-Dec-21	N
33-3	43	6	1BB	XX: +19; -22	DECLINED TESTING	20-Dec-21	N
35	42	5	1BC	XY: -21	DECLINED TESTING	23-Sep-19	N
36	45	5	2BB	XY: -21	DECLINED TESTING	9-Oct-21	Biochemical
36	45	6	1AB	XX: -15	DECLINED TESTING	9-Oct-21	Biochemical
37	40	6	2CC	XX;-16	DECLINED TESTING	9-Dec-23	N
38	38	6	1CC	TRIPLOIDY (69, XXY)	DECLINED TESTING	13-Dec-19	N
39	39	6	1BA	XY: +21; (+11, 20%)	DECLINED TESTING	18-Sep-17	N
40	36	6	1BB	XY: +16; +19	DECLINED TESTING	17-Jul-21	N
41	48	6	1CC	XX;+2;+8;-15	DECLINED TESTING	3-Dec-21	N
41	48	6	3DD	XX: +7;+9;-11;-18	DECLINED TESTING	24-Sep-23	N
41	48	6	2CC	XY;+13;+19;+21;-22	DECLINED TESTING	17-May-23	N
42	37	6	1BC	XY: +1; -12	DECLINED TESTING	22-Jan-24	N
42	37	6	1BB	XX;+1	DECLINED TESTING	22-Jan-24	N
42	37	6	1AA	XX: +15; +21	DECLINED TESTING	7-Jul-23	N
47	48	5	2CC	TRIPLOIDY (69, XXY)	DECLINED TESTING	21-Aug-24	N
50	37	6	2CC	XY: -19	DECLINED TESTING	4-Oct-24	N
51	40	6	1BB	XY: -16	DECLINED TESTING	22-Jul-24	N
52	42	7	2CC	XY: +X	DECLINED TESTING	23-Oct-24	N
					DECLINED		

Patient Number	Age	Biopsy Day
2	40	5
7	39	6
9	44	6
15	41	6
17	46	6
24	37	6
26	40	5
29	30	6
38	38	5
1	48	6
4	43	7
6	33	6
26	40	6
34	32	6
43	40.3	6
44	38.1	6
45	39.7	7
46	33.3	6
48	33.5	6
49	37.2	7

								Gestational	
Patient Number	Age	Biopsy Day	Grade	NGS PGT-A result	Reason for aneuploid transfer	FET DATE	Clinical Pregnancy outcome	Gestational age of miscarriage (w=weeks; d=days)	Follow up genetic testing
Whole Chromosome Aneuploid Embryo Transfers resulting in Miscarriage									
2	40	5	2BB	XY: +18	FRESH TRANSFER	23-Jan-18	Termination	12w4d	XY: +18
7	39	6	2BB	XX: +2; +14; +16; +18; (+9, 65%)	DECLINED TESTING	27-Oct-19	Miscarriage	5w	no
9	44	6	1BB	XY: +21	DECLINED TESTING	28-Sep-19	Miscarriage	9w	XY: +21
15	41	6	1BB	XY: +12	DECLINED TESTING	5-Oct-16	Miscarriage	6w	XY: +12
17	46	6	1BA	XY: +10	DECLINED TESTING	18-Apr-24	Miscarriage	10w	no
24	37	6	1BB	XX: +15	DECLINED TESTING	1-Apr-24	Miscarriage	5w3d	no
26	40	5	1BB	XY: +16	DECLINED TESTING	16-Dec-20	Miscarriage	7w6d	XY: +16
29	30	6	1BA	XY: +9	DECLINED TESTING	26-Jan-21	Miscarriage	6w1d	XY: +9
38	38	5	1CB	XY: +22	DECLINED TESTING	28-Jul-19	Miscarriage	9w4d	no
Segmental Aneuploid Embryo Transfers									
1	48	6	1BB	XX: -3q22.1-q29|65.3Mb	DECLINED TESTING	20-Jan-18	Live birth		no
4	43	7	2DD	XY: -2p23.2-p11.2|29.9Mb; -11q14.3-q22.3|15.1Mb	DECLINED TESTING	21-Apr-23	N		
6	33	6	1BC	XX: -2q37.1-q37.3|11.3Mb	DECLINED TESTING	21-Feb-23	N		
26	40	6	1CB	XX: -7q32.1-q36.3|30.36Mb	DECLINED TESTING	24-May-21	N		
34	32	6	1BB	XY: -18q21.1-q23|33.1Mb	DECLINED TESTING	19-Mar-24	N		
43	40.3	6	1CC	XY: -1p36.33-p36.22|12.1Mb; +1p36.22-p21.1|94.6Mb	DECLINED TESTING	19-Oct-23	Live bi [No Title]		no
44	38.1	6	1CC	XX: -1q|	DECLINED TESTING	17-Feb-23	Miscarriage	7w6d	no
45	39.7	7	3DD	XY: +Xp22.33|18Mb	DECLINED TESTING	8-Dec-22	N		
46	33.3	6	1BC	XX: -2q37.1-q37.3|11.3Mb	DECLINED TESTING	21-Feb-23	N		
48	33.5	6	2AB	XY: -10q23.1-q26.3|52Mb	DECLINED TESTING	31-Jul-24	N		
49	37.2	7	1CD	XX: -2p|	DECLINED TESTING	10-Sep-24	Ongoing pregnancy		