

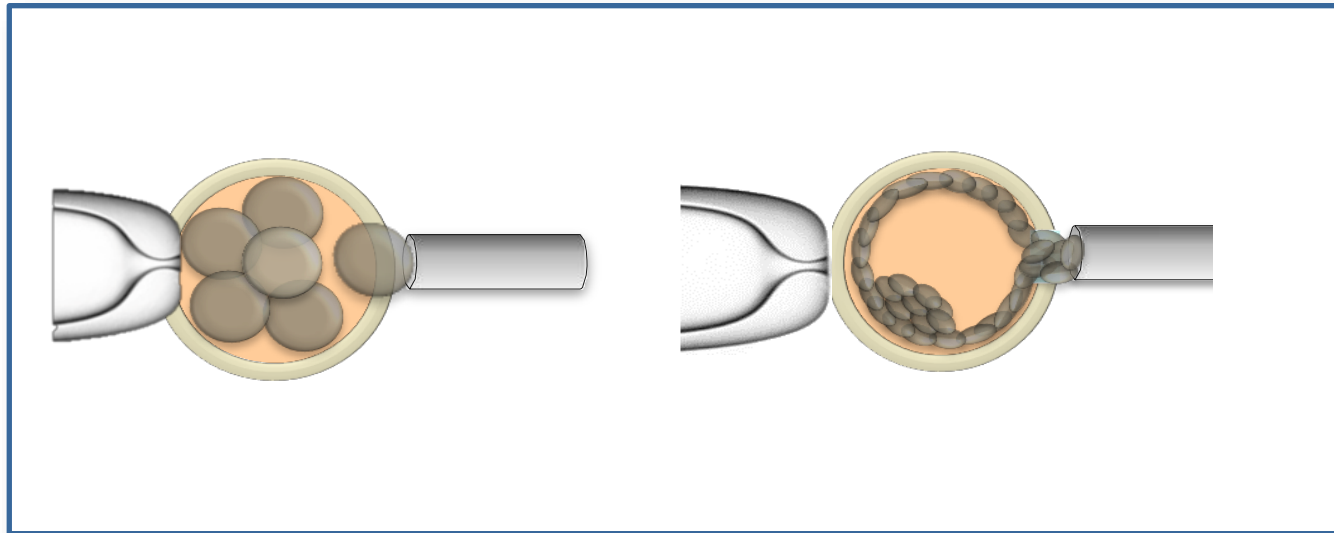


Análise genética embrionária

Edson Borges Jr.

Diagnóstico Genético Pré-Implantacional

Retirada de uma ou duas células de um pré-embrião
no dia +3 (4 - 8 células) / 5 células do dia +5 (120 células)
durante seu desenvolvimento laboratorial para análise genética



FERTILITY

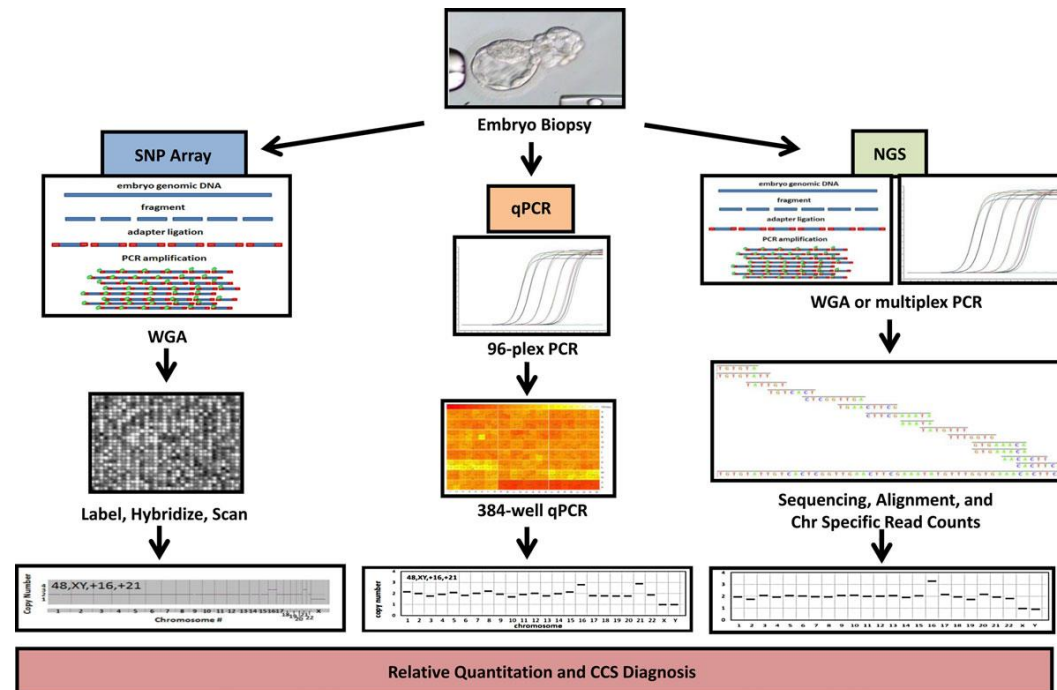
Diagnóstico Genético Pré-Implantacional



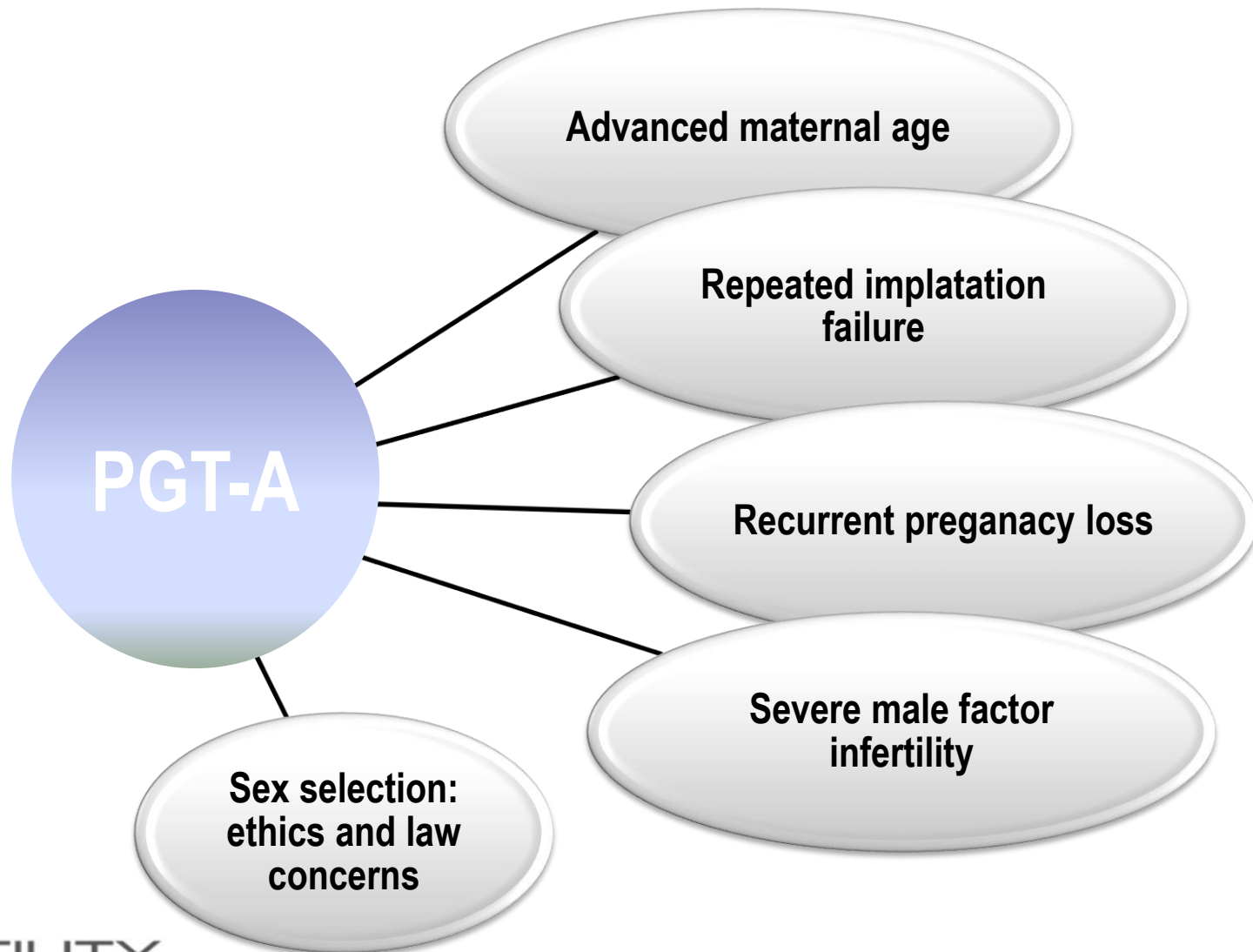
- Aberrações cromossômicas numéricas: **PGT-A**
- Aberrações cromossômicas estruturais: **PGT-SR**
- Alterações monogênicas (*single genes defects*): **PGT-M**
- *Human leukocyte antigen* (antígeno de histocompatibilidade): **HLA**
- Alterações poligênicas: **PGT-P**

Diagnóstico Genético Pré-Implantacional

- **arrayCGH** (comparative genome hybridization),
- **arraySNP** (single nucleotide polymorphism),
- **qPCR** (real-time quantitative polymerase-chain-reaction)
- **NGS** (next-generation sequencing)

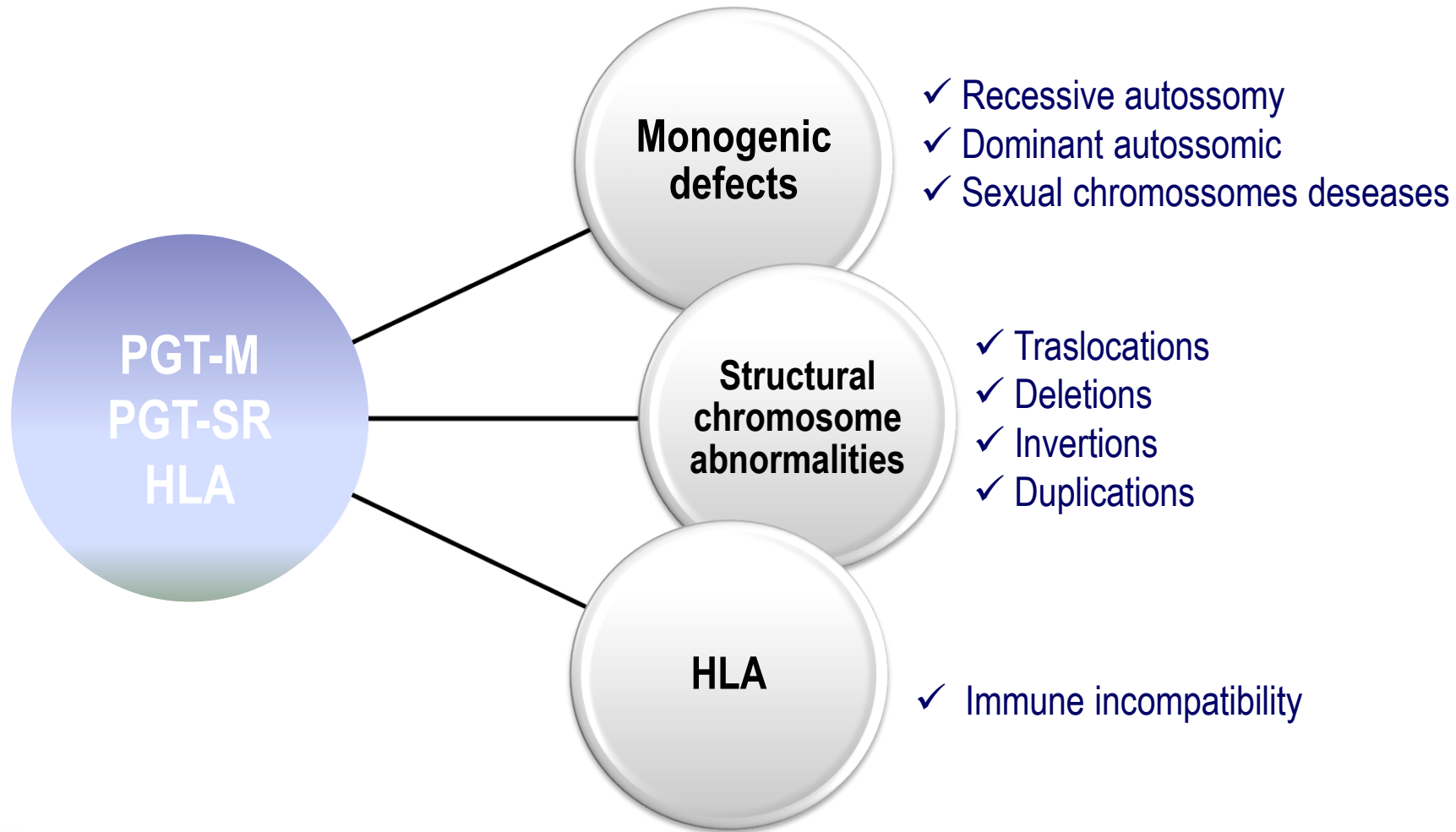


Diagnóstico Genético Pré-Implantacional

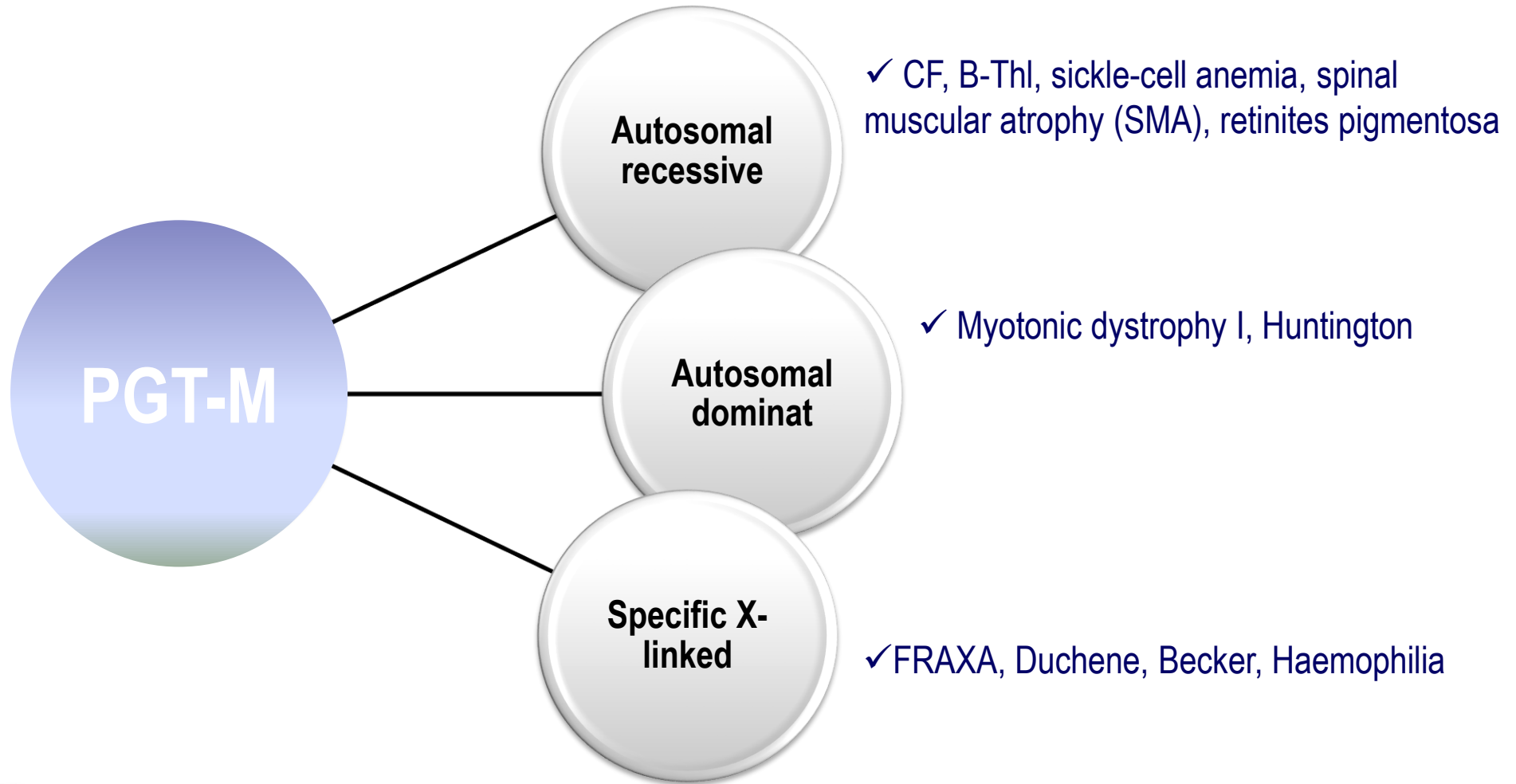


FERTILITY

Diagnóstico Genético Pré-Implantacional



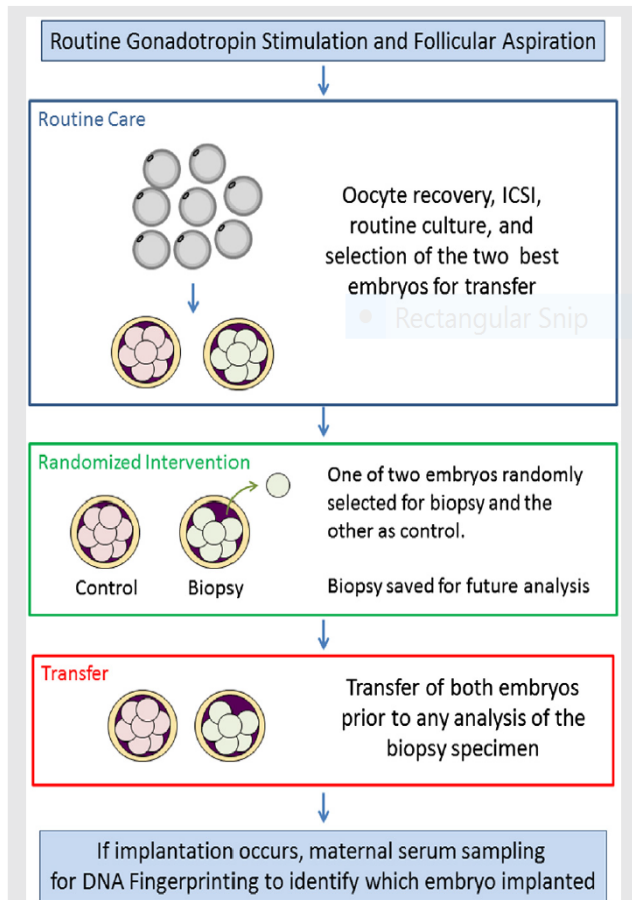
Diagnóstico Genético Pré-Implantacional





FERTILITY

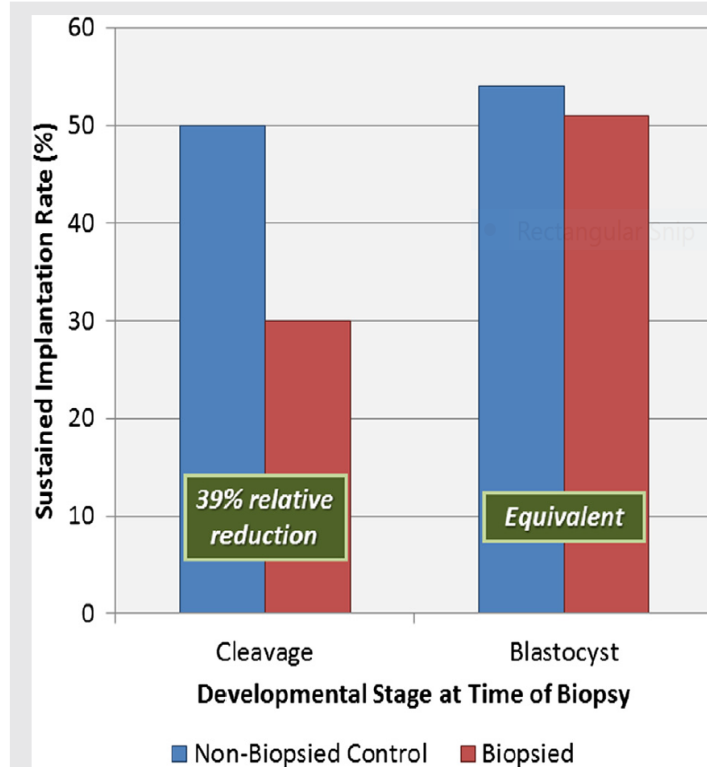
FIGURE 1



Study design to assess the impact of biopsy at the cleavage stage (day 3) on embryonic reproductive potential. The randomized paired experimental design allowed each patient to serve as her own control. An equivalent methodology was used to determine if trophectoderm biopsy at the blastocyst stage affected embryonic potential. ICSI = intracytoplasmic sperm injection.

Scott. Cleavage-stage biopsy is harmful. Fertil Steril 2013.

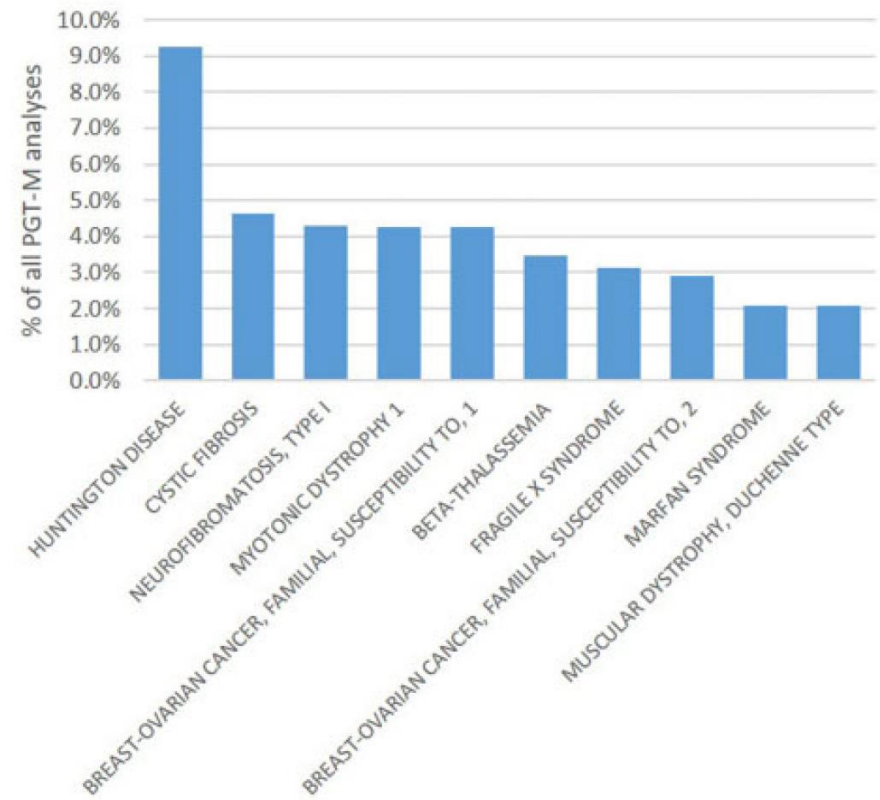
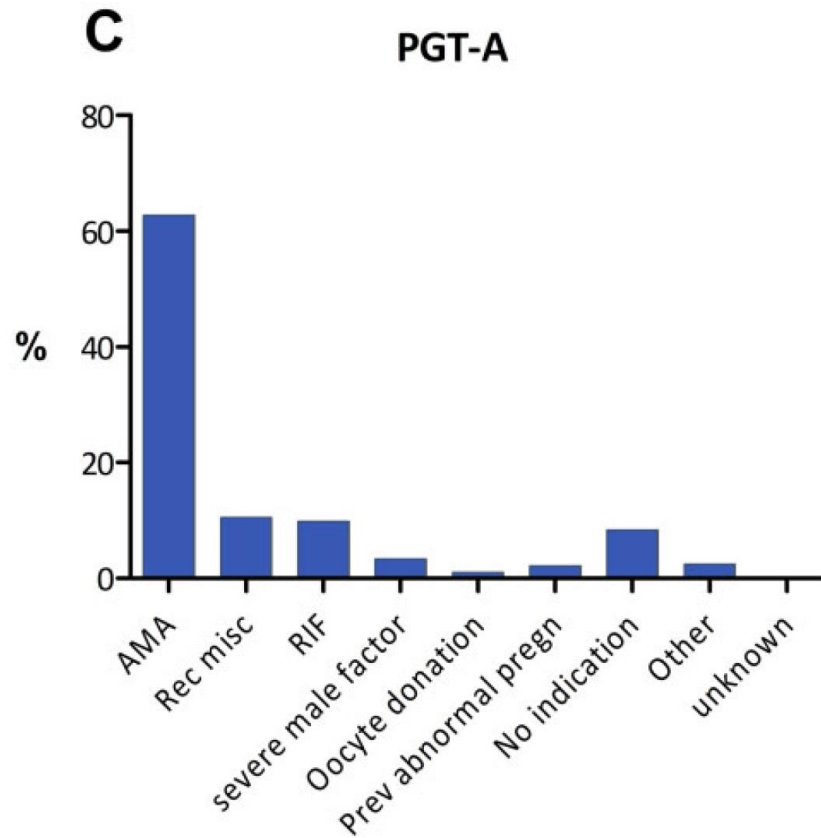
FIGURE 3



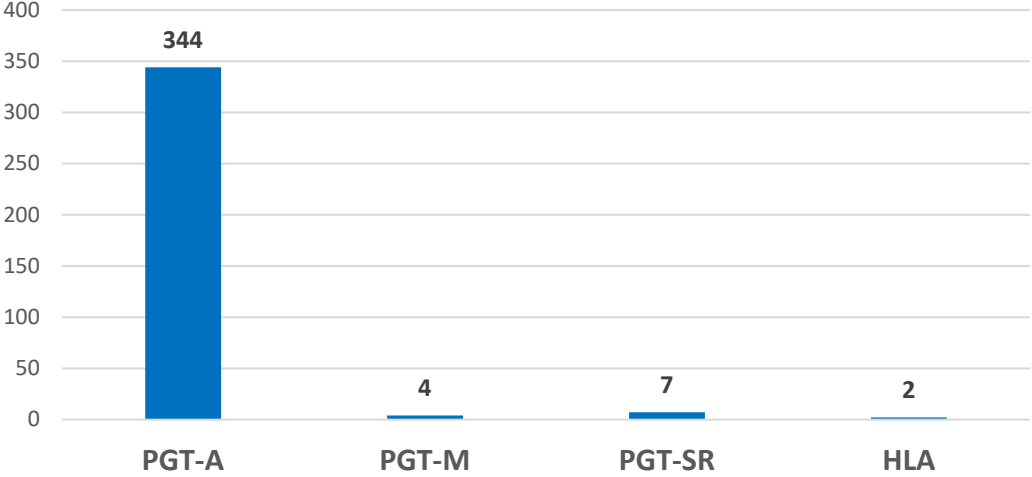
Implantation rates following a randomized paired analysis of the effects of cleavage- and blastocyst-stage biopsies on embryo reproductive potential. Sustained implantation and delivery of the biopsied embryo were significantly reduced compared with its control sibling when biopsy was performed on day 3 at the cleavage stage (McNemar chi-square: $P < .03$). A similar paired analysis demonstrated that the developmental potential of embryos undergoing trophectoderm biopsy at the blastocyst stage was equivalent to the nonbiopsied control siblings.

Scott. Cleavage-stage biopsy is harmful. Fertil Steril 2013.

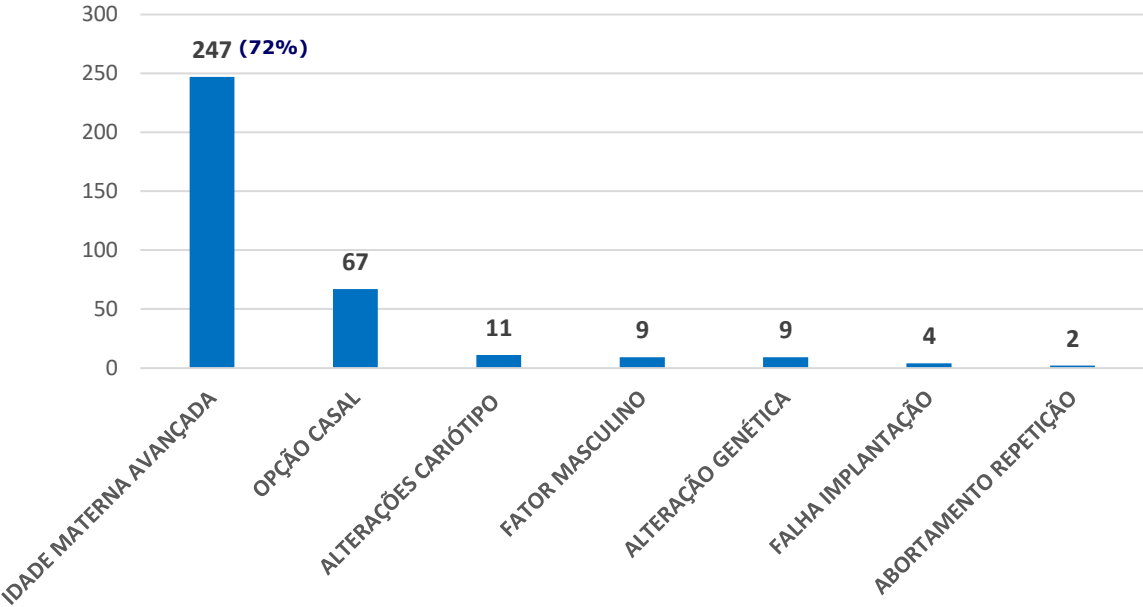
ESHRE PGT Consortium data collection XIX–XX: PGT analyses from 2016 to 2017[†]



Fertility 2021

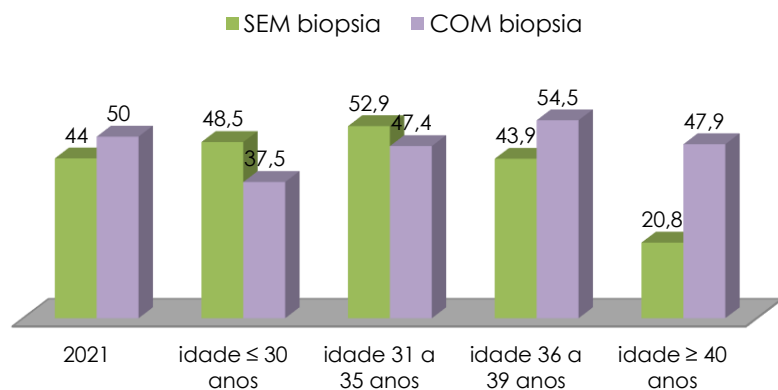


PGT-A Fertility 2021

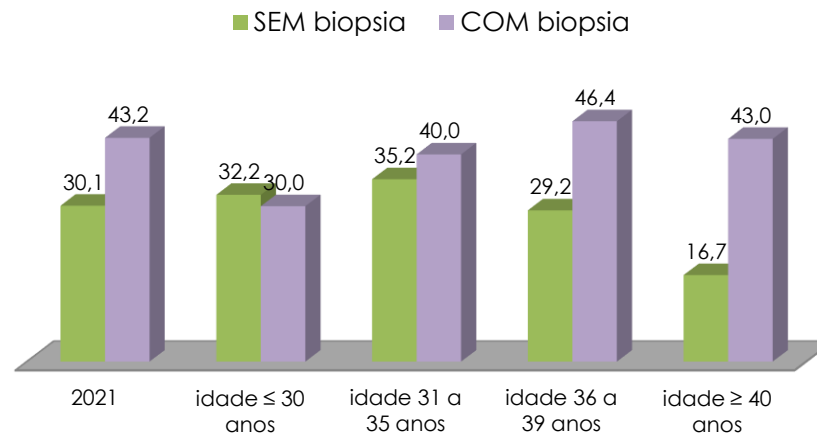


		Embrões sem biópsia					Embrões com biópsia				
		n° ciclos transf	tx gest bioquímica	tx gest clínica	tx aborto	tx implantação	n° ciclos transf	tx gest bioquímica	tx gest clínica	tx aborto	tx implantação
idade	2021	425	49,2	44	11,2	30,1	224	56,3	50	13,4	43,2
	idade ≤ 30 anos	68	52,9	48,5	6,1	32,2	8	37,5	37,5	0,0	30,0
	idade 31 a 35 anos	153	56,2	52,9	11,1	35,2	57	56,1	47,4	7,4	40,0
	idade 36 a 39 anos	132	49,2	43,9	13,8	29,2	88	58,0	54,5	12,5	46,4
	idade ≥ 40 anos	72	30,6	20,8	13,3	16,7	71	56,3	47,9	20,6	43,0

Tx de gestação clínica /ciclo transf (%)



Tx de implantação (%)



SAÚDE

EDITORA



Abril

premiossaude@abril.com.br

► Finalistas

Saúde Mental e Emocional

Saúde e Atividade Física

Saúde Bucal

Saúde e Nutrição

Saúde e Prevenção

Saúde da Criança

Instituição do Ano



FERTILITY

Diagnóstico pré-implantacional para Beta-talassemia combinado com compatibilidade HLA: nascimento do primeiro "irmão salvador" após seleção embrionária no Brasil

Autores: Ciro Dresch Martinhago e Edson Borges Junior

Instituições: Chromosome Medicina Genômica e Fertility - Centro de Fertilização Assistida

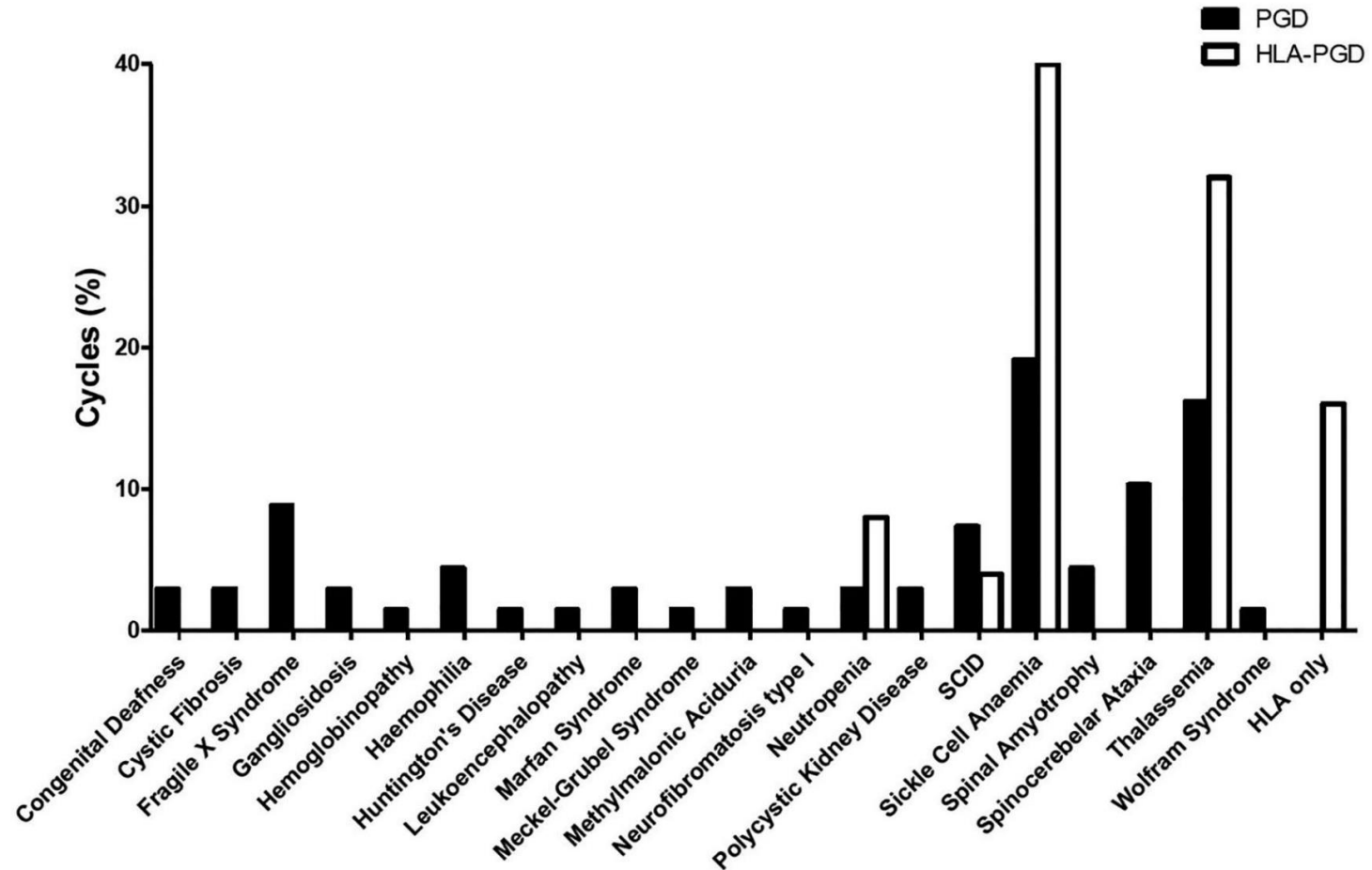


Preimplantation genetic testing for monogenic diseases: a Brazilian IVF centre experience

Bianca Ferrarini Zanetti^{1,2}, Daniela Paes de Almeida Ferreira Braga¹, Matheus de Castro Azevedo¹, Amanda Souza Setti^{1,2}, Rita Cássia Sávio Figueira¹, Assumpto Iaconelli Jr.^{1,2}, Edson Borges Jr.^{1,2}

¹Fertility - Medical Group, São Paulo, SP - Brazil

²Instituto Sapientiae - Centro de Estudos e Pesquisa em Reprodução Humana Assistida, São Paulo, SP - Brazil



Diagnóstico Genético Pré-Implantacional

www.nature.com/scientificreports

SCIENTIFIC
REPORTS

nature research

Corrected: Author Correction

OPEN Genomic Prediction of 16 Complex Disease Risks Including Heart Attack, Diabetes, Breast and Prostate Cancer

Louis Lello¹, Timothy G. Raben¹, Soke Yuen Yong¹, Laurent C. A. M. Tellier^{2,3} & Stephen D. H. Hsu^{1,2,3}

❖ 2020: PGT-P: Hypothyroidism, (Resistant) Hypertension, Type 1 and 2 Diabetes, Breast Cancer, Prostate Cancer, Testicular Cancer, Gallstones, Glaucoma, Gout, Atrial Fibrillation, High Cholesterol, Asthma, Basal Cell Carcinoma, Malignant Melanoma, Heart Attack, Schizophrenia, Autism.

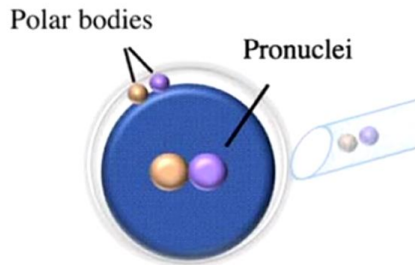
Genetic Disease Prevalence



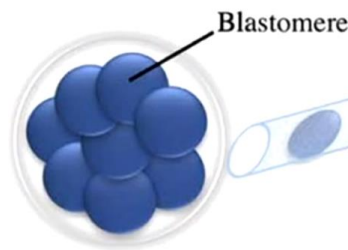
FERTILITY

BIOPSY METHODS

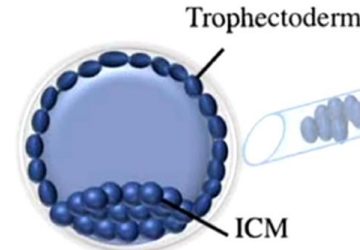
Preimplantation development →



Pronuclear stage

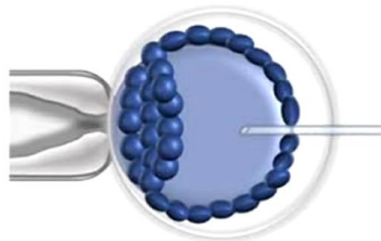


Cleavage stage

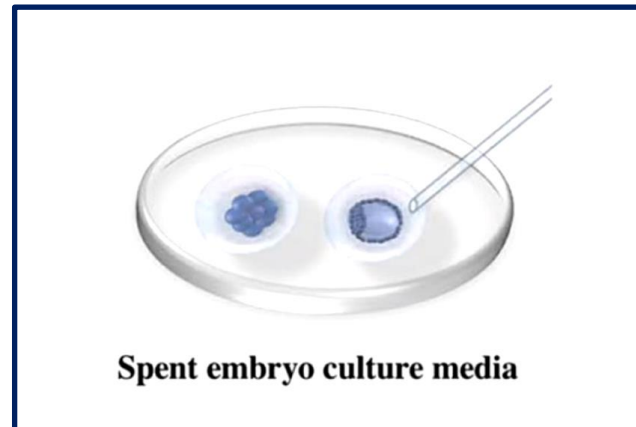


Blastocyst stage

Traditional cell-biopsy methods



'Blastocentesis'

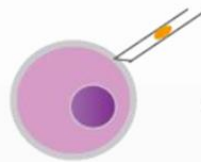


Spent embryo culture media

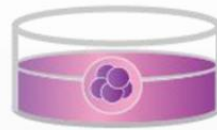
Minimally/non-invasive approaches



Oocyte



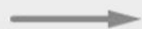
ICSI



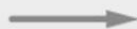
**D5-D6
Blastocyst**



**Culture Medium
Collection**



**NICS™ Sample
Preparation**



**Next Generation
Sequencing**



**Result
Reporting**



FERTILITY

Resultado Geral da validação NiPGTA

Concordância clínica					
	NICS Yikon (Ion + Illumina)		NICS Reproseq PGS		PGT-A
	vs. PGT-A	vs. Embrião	vs. PGT-A	vs. Embrião	vs. Embrião
TOTAL	51	15	26	16	20
CONCORDANTE	30/51 (59%)	11/15 (73%)	18/26 (69%)	13/16 (81%)	15/20 (75%)
DISCORDANTE	21/51 (41%)	4/15 (27%)	8/26 (31%)	3/16 (19%)	5/20 (15%)
Falha de amplificação (DISCORDANTE)	2/51 (4%)	0/15 (0%)	4/26 (15%)	1/16 (6%)	0/20 (0%)

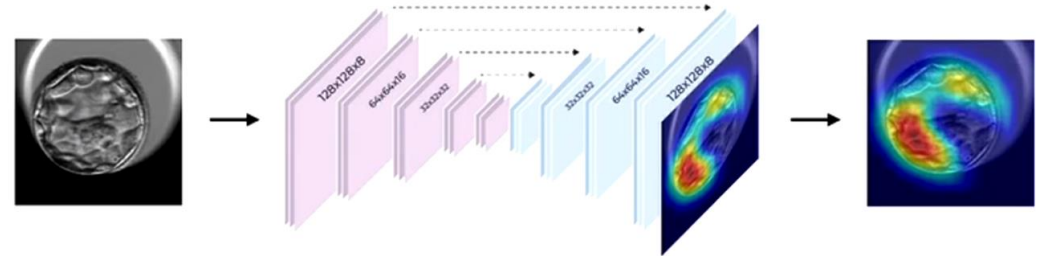
Obs: para os cálculos de concordância foram excluídos os casos testados com protocolo alternativo de WGA.

A taxa de concordância entre PGTA e meio de cultivo foi de 69%.

São necessários mais estudos para ampliar essa taxa de concordância e trazer confiabilidade para o teste não invasivo com resultados robustos em taxas semelhantes ao diagnóstico de PGTA.

Espera-se encontrar taxas de concordância de no mínimo 90% para levar o teste para diagnóstico.

● Non-invasive approaches to distinguish between euploid and aneuploid embryos: **ni-AI-PGT-A**



> *Reprod Biomed Online*. 2020 Oct;41(4):585-593. doi: 10.1016/j.rb. 70% precision

Embryo Ranking Intelligent Classification Algorithm (ERICA): artificial intelligence clinical assistant
 predic > *J Assist Reprod Genet*. 2021 Jul;38(7):1655-1663. doi: 10.1007/s10815-021-02228-8.
 Epub 2021 May 22.

Alejandro C
 Andrew J D

**End-to-end deep learning for recognition o
 status using**

Chun-I Lee ^{1 2 3}, Yan-R
 Wei-Lin Zheng ⁴, Wen-T

**P-203 Applying artificial intelligence for ploidy
 prediction: The concentration of IL-6 in spent
 culture medium, blastocyst morpholo
 and embryo morphokinetics as variab
 consideration** [Get access >](#)

B Aparicio Ruiz, L Bori, E Paya, M A Valera, A Quiñero, F Domin
Human Reproduction, Volume 36, Issue Supplement_1, July 202

65% precision

> *Fertil Steril*. 2022 Apr;117(4):738-746. doi: 10.1016/j.fertnstert.

**Machine learning for pred
 human embryos: in search
 model and predictive feat**

Stefanie De Gheselle ¹, Céline Jacques ², Jérôme
 Ilse De Croo ⁴, Cristina Hickman ², Kelly Tilleman

72% precision

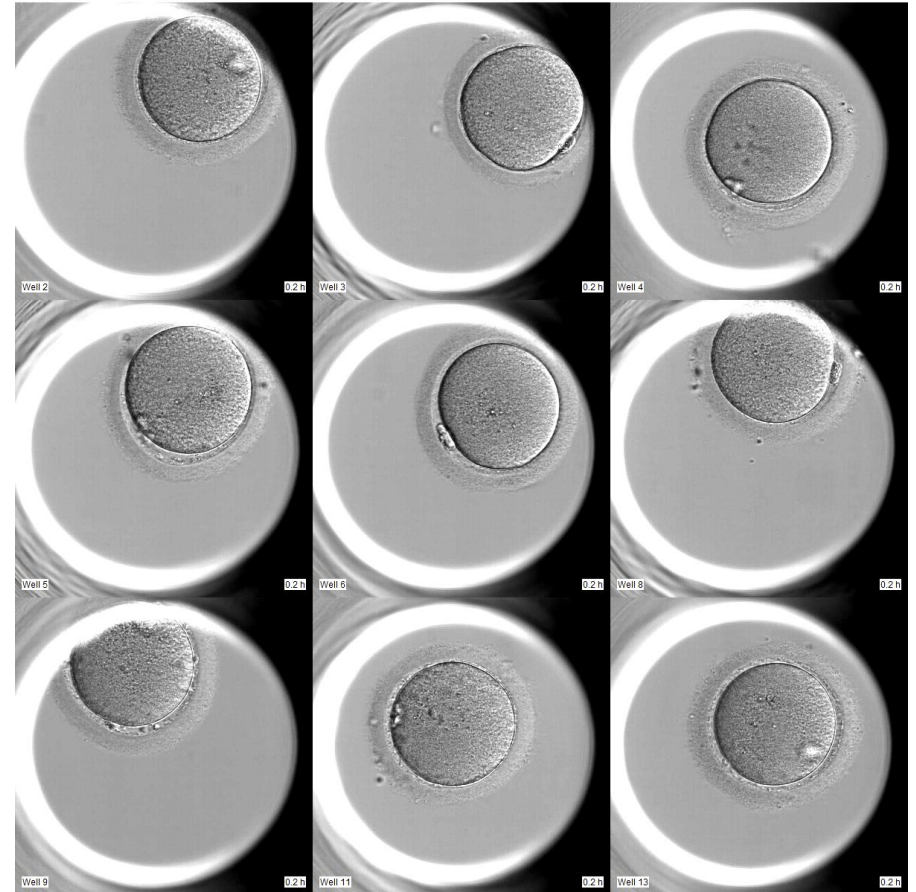
> *Reprod Biol Endocrinol*. 2021 Dec 13;19(1):185. doi: 10.1186/s

**An artificial intelligence model (euploid prediction
 algorithm) can predict embryo ploidy status based on
 time-lapse data**

Bo Huang ¹, Wei Tan ¹, Zhou Li ², Lei Jin ³

78,5% precision

Embryoscope



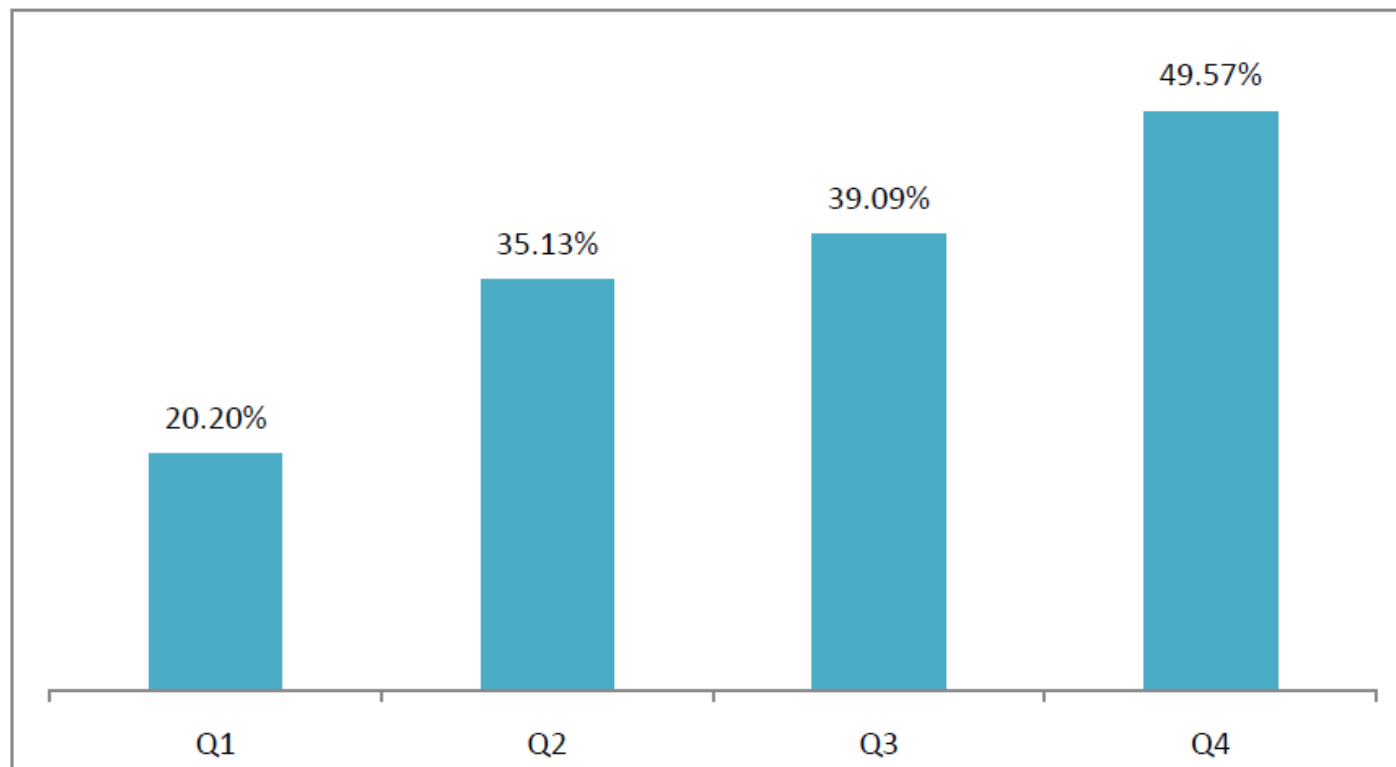


Figure 2: Distribution of the percentage of euploid embryos into the KIDScore D 5 categories, Q1 ≤ 3.9 , Q2, between 4 and 5.6, Q3 between 5.7 and 7.5, and Q4 ≥ 7.6



Borges E. et al

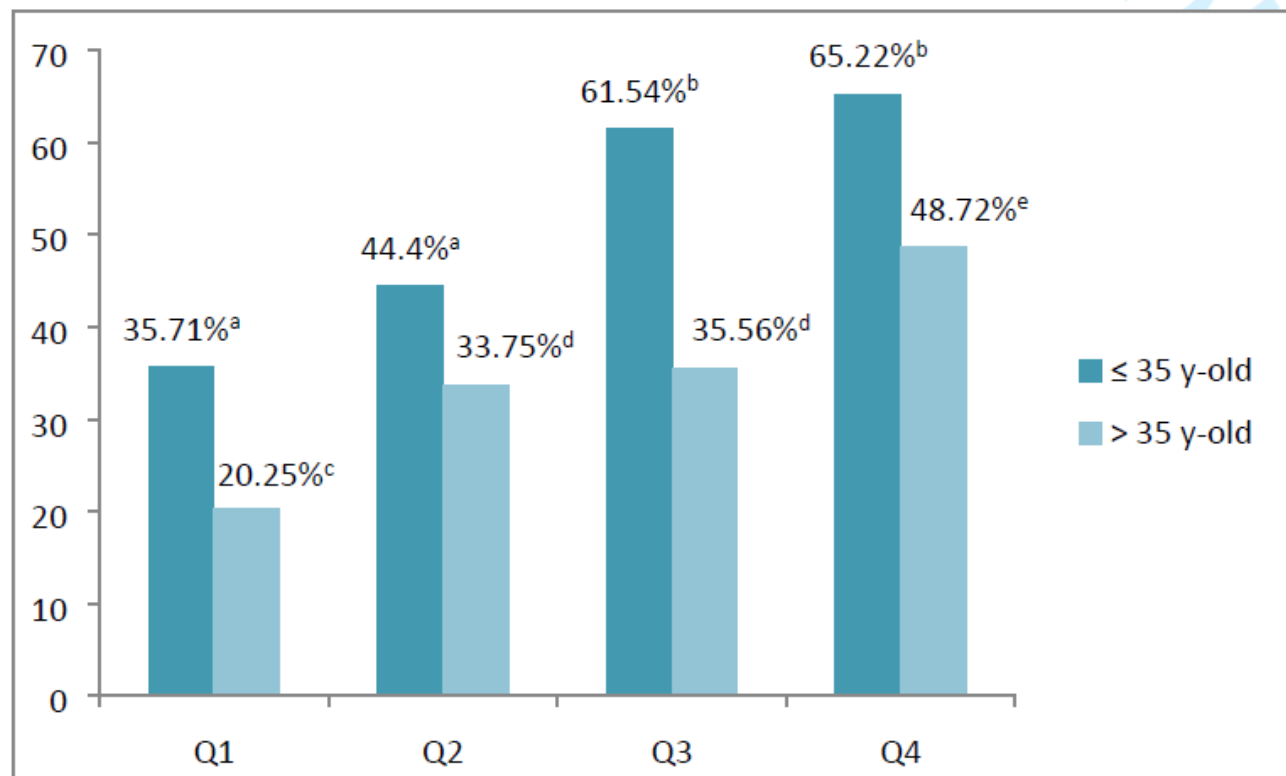


Figure 3: The distribution of the chance of being euploid according with the KIDScore D 5 category: Q1 ≤ 3.9 , Q2 between 4 and 5.6, Q3 between 5.7 and 7.5, and Q4 ≥ 7.6 . $a \neq b \neq c \neq d \neq e$.



Borges E. et al



FERTILITY



Direção

Assumpto Iaconelli Jr.
Edson Borges Jr.



Pesquisa e Educação

Amanda Setti
Christina Morishima
Daniela Braga
Joana Nogueires Simas
Lorrana de Souza Anjos



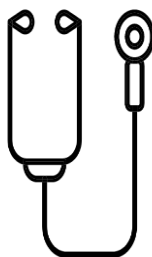
Psicologia

Rose M. Melamed



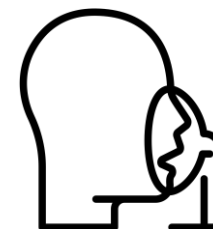
Laboratório de FIV

Kelly C. Pinheiro
Livia Silvia Vingris
Patrícia Guilherme
Rafaela Ferrazi Maluf
Tatiana Nunes de Melo



Corpo Clínico

Barbara Brigati
Carla Iaconelli
Edward Carrilho
Fernanda Montenegro
Fábio Biaggioni Lopes
Graziela C. C. Carvalho
Mauro Bibancos
Natalia G. Tannous
Paula Ferreiro Vieira
Mario Firmino



Anestesiologia

Marcelo Torres e
Equipe



Laboratório de Andrologia

Rodrigo Rosa Provenza
Thais de Almeida Silva Ferreira



Nutrição

Gabriela Halpern

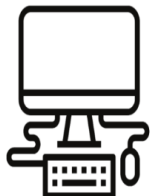


FERTILITY



Enfermagem

Ana Maria Domingos França
Gabriela Lima Almeida
Larissa Rodrigues Gonçalves
Maria Regina S. da Silva
Vera Lucia Alves



Informática

Marcelo Alexandre Baptista



Farmácia

Maria das N. Fernandes



Suporte

Francilene Faustino Lopes
Francisco Junior M. Anjos
Hugo Oliveira dos Reis
Janaína Gomes Pinho da Silva
Katia Rodrigues
Leidivânia Cerqueira Alves
Leonardo S. Lopes
Lohany Gabrielly Lira
Lucácio de Souza Anjos
Marcos Vinícius de Sousa
Simone de S. Carvalho
Zenilda da Conceição Souza



Administração

Margaret Meira
Fabiana Garcia

Obrigado!

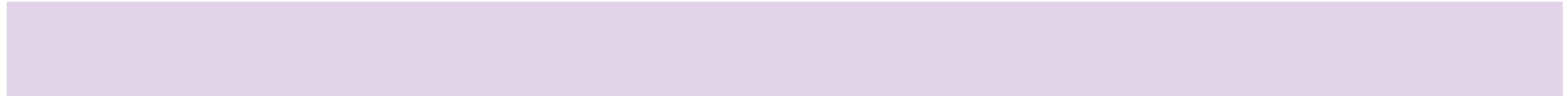
Dr. Edson Borges Jr.

www.fertility.com.br

E-mail: edson@fertility.com.br



FERTILITY
MEDICAL GROUP



FERTILITY