Clinical application of PGT-M for HLA-typing in Latin America

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Conflicts of interest

Scientific Advisor

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INTRODUCTION

PGT-M for Human Leukocyte Antigen (HLA)-typing enables the selection of healthy and HLA-compatible embryos with an affected sibling. This approach provides access to stem cell therapies, such as hematopoietic stem cell transplantation. Couples must follow IVF treatments, for embryo testing.

PGT-M only for HLA-typing:
25% of probability for an embryo HLA-matched.

PGT-M for HLA-typing with the exclusion of monogenic diseases:
The genetic chance of an HLA-matched and healthy embryo drops.
18,8% for autosomal recessive o x-linked disease.
12,5% for autosomal dominant.

Kakourou et al., 2018.

INTRODUCTION

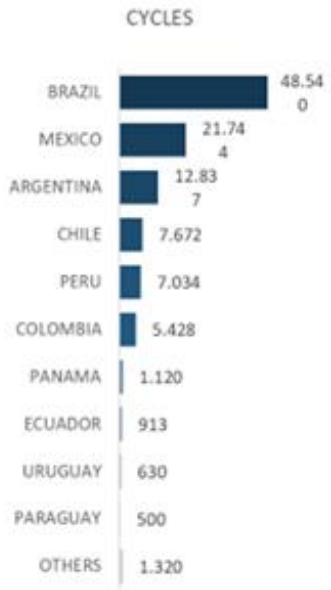
About Latin America

No specific regulations for PGD.

Embryo biopsy was only prohibited in Costa Rica.

Biopsies are always performed at blastocyst stage.

TOTAL OF IVF CYCLES BY COUNTRY



OBJECTIVE

This study aimed to review PGT-M for HLA-typing cases from Latin America, focusing on the clinical success concerning the number of embryos available for transfer.

Retrospective study: PGT-M/HLA-typing cases performed in Latin America between 2017-2024.

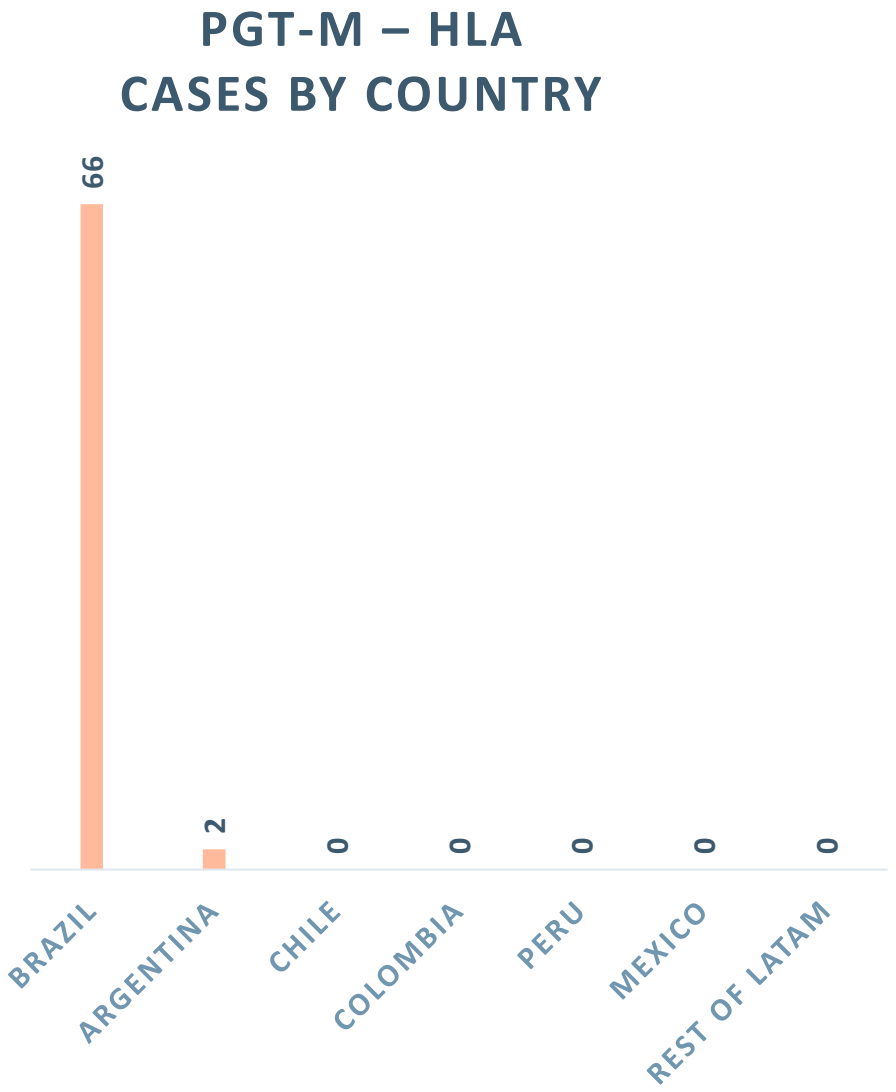
RESULTS IN LATIN AMERICA

Total of 1150 PGT-M cases

From **Brazil**: 66 / 815 PGT-M cases (**8,1%**).

From **Argentina**: 2 / 129 PGT-M cases (**1,6%**).

From rest of **LATAM**: 0 / 206 PGT-M cases (**0%**)

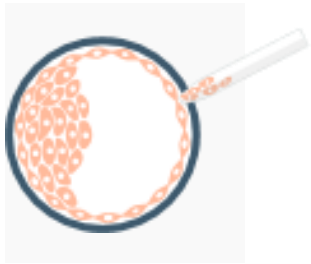


RESULTS IN LATIN AMERICA

Cases from Brazil and Argentina:

68 PGT-M/HLA-typing cycles (7,2% from total PGT-M).

44 couples - 469 blastocysts



Maternal age: 34.9 years ± 4,8

Indirect HLA haplotyping (by STR markers).

PGT-A was also added.

RESULTS IN LATIN AMERICA

Only HLA-typing

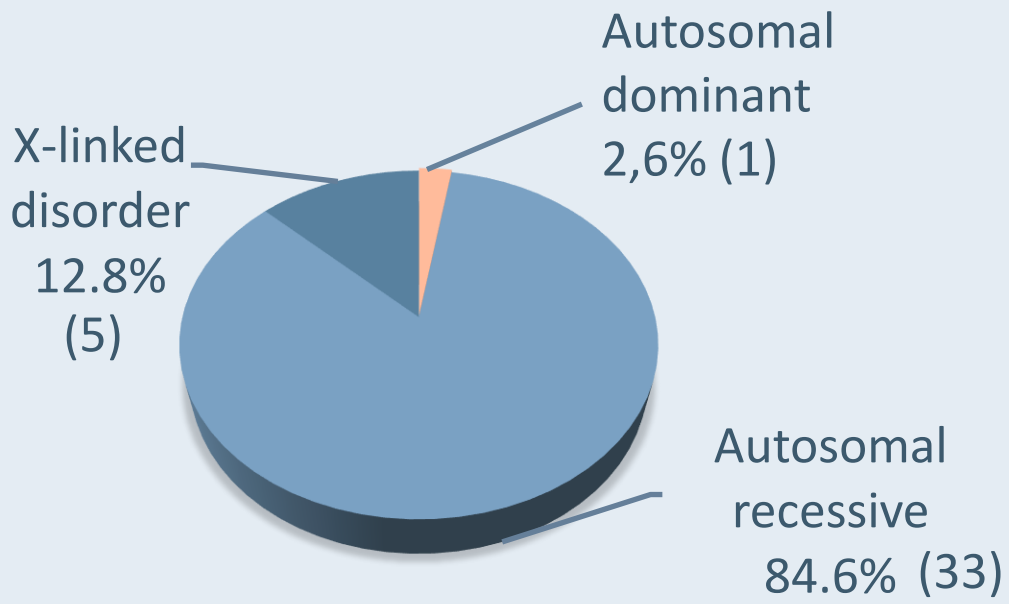
5 couples (11.3%): 6 PGT-M cycles.
For leukemia diseases.

HLA-typing + monogenic diseases

39 couples (88.7%) - 62 PGT-M cycles.

Autosomal recessive - 94% HBB gene:

About 4% of Brazil's population has the sickle cell trait, and 60.000 to 100.000 have SCD.



RESULTS IN LATIN AMERICA

	HLA only	HLA with monogenic disorder	Autosomal dominant	Autosomal recessive	X-linked disorder
Number of embryos analyzed	**35**	**434**	1	388	45
HLA-matched	**22.9% (8/35)**	25.8% (112/434)	1	100	11
Unaffected for the disease	-	**17.2% (75/434)**	100% (1/1)	16.8% (65/388)	20% (9/45)
Affected for the disease	-	37	-	35	2
Non-HLA matched embryos	26	304	-	273	31
Unaffected for the disease	-	200	-	178	22
Affected for the disease	-	104	-	95	9
Incomplete/inconclusive diagnosis	1	18	-	15	3
TRANSFERABLE EMBRYOS (HLA-matched (unaffected) and euploid)	**8.6% (3/35)**	**11.5% (50/434)**	**1**	**10.8% (42/388)**	**15.5% (7/45)**
Maternal age	**37.4 ± 2.5**	**34.6 ± 5,0**	**26**	**35.2 ± 4,8**	**32.5 ± 5,0**

RESULTS IN LATIN AMERICA

Mean of embryos analyzed per couple: 10.6.

Couples performed from **1 to 6 PGT-M** (mean: 2).

A total of 53 embryos (11.3%) were available for transfer:

Publication:	Embryos suitable for transfer:
Kakourou et al., 2018	15.6%
De Rycke et al., 2020	16,5%
Our results	11,3% (17,7% without PGT-A)

30 couples (68.2%) had at least one embryo for transfer.
14 couples (31.8%) did not achieve an embryo transfer and gave up their IVF treatment.

RESULTS IN LATIN AMERICA - Success story

Liz, que sofre de anemia falciforme, ao lado de sua irmã, Isa
Imagem: Arquivo pessoal

Anemia falciforme: Liz receberá medula da irmã para se curar da doença

Giulia Granchi
Do VivaBem, em São Paulo
19/12/2021 04h00

SICKLE CELL ANEMIA:

In Brazil, 3.500 children are born with the disease each year: 1 baby for every 1.000 births.

The 20% of children do not reach the age of 5.

The only cure is through a bone marrow transplant.

Girl diagnosed with **sickle cell anemia** at 4 months old:

In April 2017, the couple saw a news report and discover that embryo selection was possible.

2019 - The couple proceed with an IVF treatment:

3 PGT-M cycles – 18 embryos (3 embryos for transfer)

1° embryo transfer: gestational loss.

2° embryo transfer: healthy and HLA compatible baby girl.

2022 - the affected daughter received the treatment.

CONCLUSIONS

PGT-M could be valuable for achieving an HLA-matched pregnancy with an affected offspring.

The first PGT-M case for HLA was reported in 2001 (Verlinsky et al., 2021).

However, there are limited cases in Latin America.

- Lack of awareness regarding its availability in these countries, among general population and medical community.
- Complex procedure. The need for many embryos to ensure a suitable match.
- Economical issues.
- Ethical issues.

Thank you for your attention

Daniela Lorenzi[1], Martina Di Bastiano[1], Larissa Antunes[2], Amanda Shinzato[2], Virginia Regla[2], Susana Joya[2], Taccyanna Mikulski Ali[2], Paula Queiroz Estrada[2], Camila Dantas[2], Bruno Copreski[2], Sebastian Salinas[1], Marcia Riboldi2, José Antonio Martinez-Conejero[3], Ana Cervero[3] Carmen Rubio[3].

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