



IPOPI
LATIN AMERICAN
PID PATIENTS'
MEETING

OCTOBER 19-20, 2023
MEXICO CITY, MEXICO

an **IPOPI** event

SESIÓN 6

COLLABORATION



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IPOPI
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an IPOPI event

Diagnóstico, tratamientos y cuidados de las IDPs

PID diagnosis, treatments and care

Moderadora: Roberta Anido de Pena

Moderator: Roberta Anido de Pena

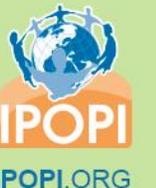
SESIÓN 6

COLLABORATION



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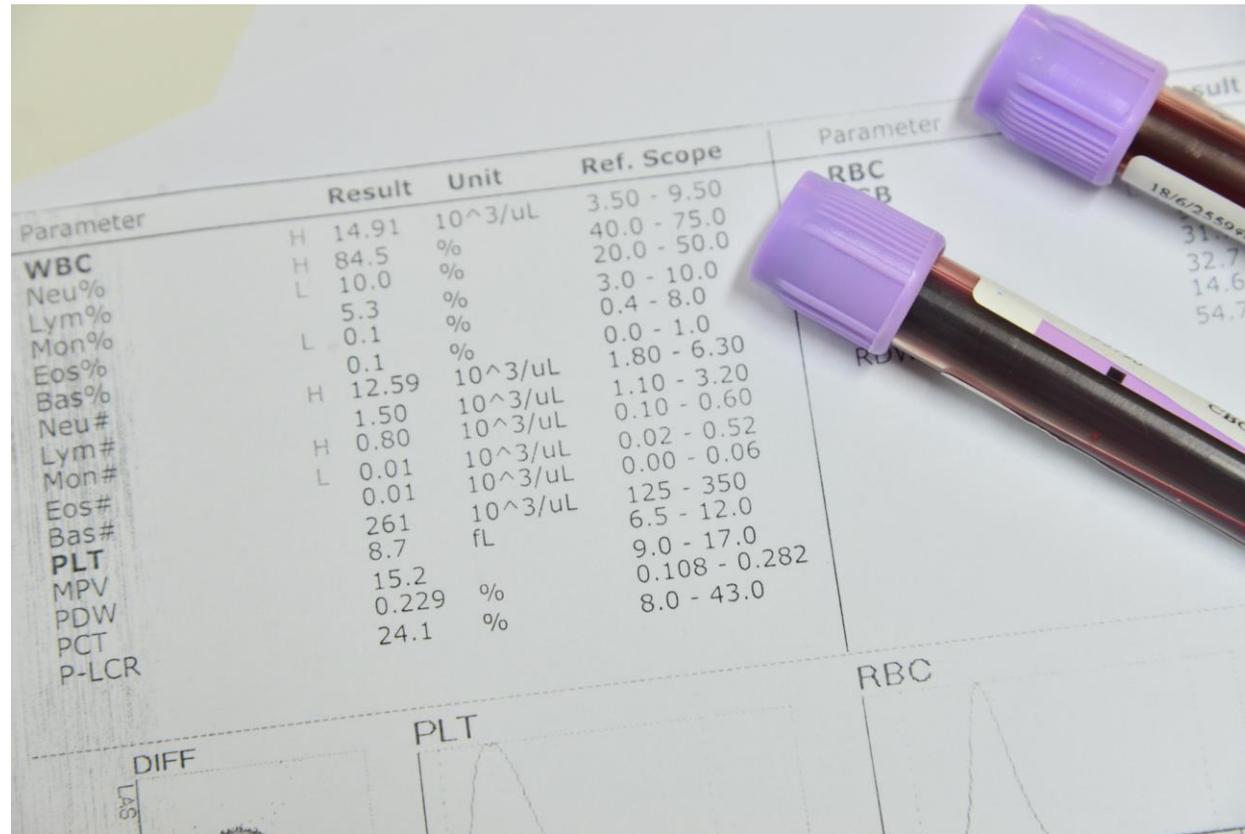
Diagnóstico: de lo básico a la genética

Essential diagnosis list

Dra Carolina Prando, Brasil
Dr Carolina Prano, Brazil

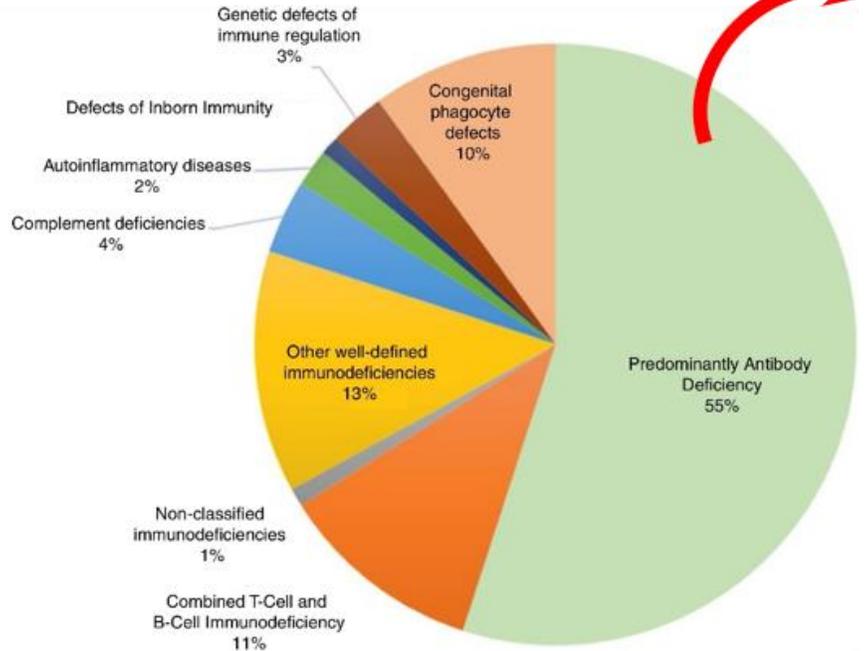






Parameter	Result	Unit	Ref. Scope
WBC	H 14.91	$10^3/uL$	3.50 - 9.50
Neu%	H 84.5	%	40.0 - 75.0
Lym%	L 10.0	%	20.0 - 50.0
Mon%	5.3	%	3.0 - 10.0
Eos%	L 0.1	%	0.4 - 8.0
Bas%	0.1	%	0.0 - 1.0
Neu#	H 12.59	$10^3/uL$	1.80 - 6.30
Lym#	1.50	$10^3/uL$	1.10 - 3.20
Mon#	H 0.80	$10^3/uL$	0.10 - 0.60
Eos#	L 0.01	$10^3/uL$	0.02 - 0.52
Bas#	0.01	$10^3/uL$	0.00 - 0.06
PLT	261	$10^3/uL$	125 - 350
MPV	8.7	fL	6.5 - 12.0
PDW	15.2	%	9.0 - 17.0
PCT	0.229	%	0.108 - 0.282
P-LCR	24.1	%	8.0 - 43.0

Conteo sanguíneo complete / hemograma



IgG, IgM, IgA, IgE; IgG subclasses

Respuesta de la IgG a las vacunas

Isohemaglutinas IgM anti-A/anti-B

Imagem:
Douglas Paes Barreto et al. *Jornal de Pediatria*: 2020.
Online 22 de novembro.

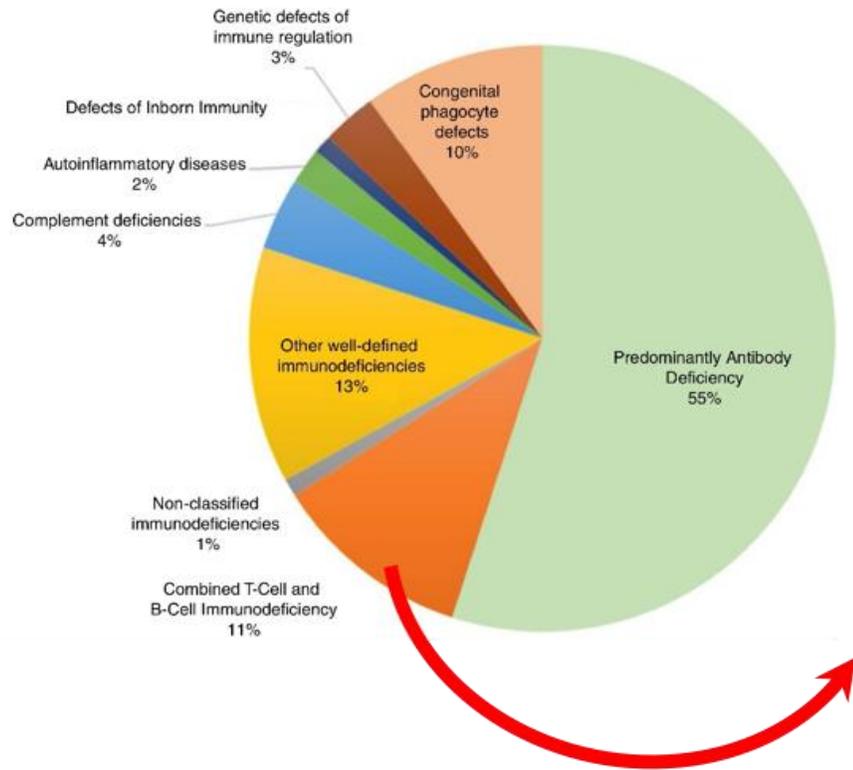
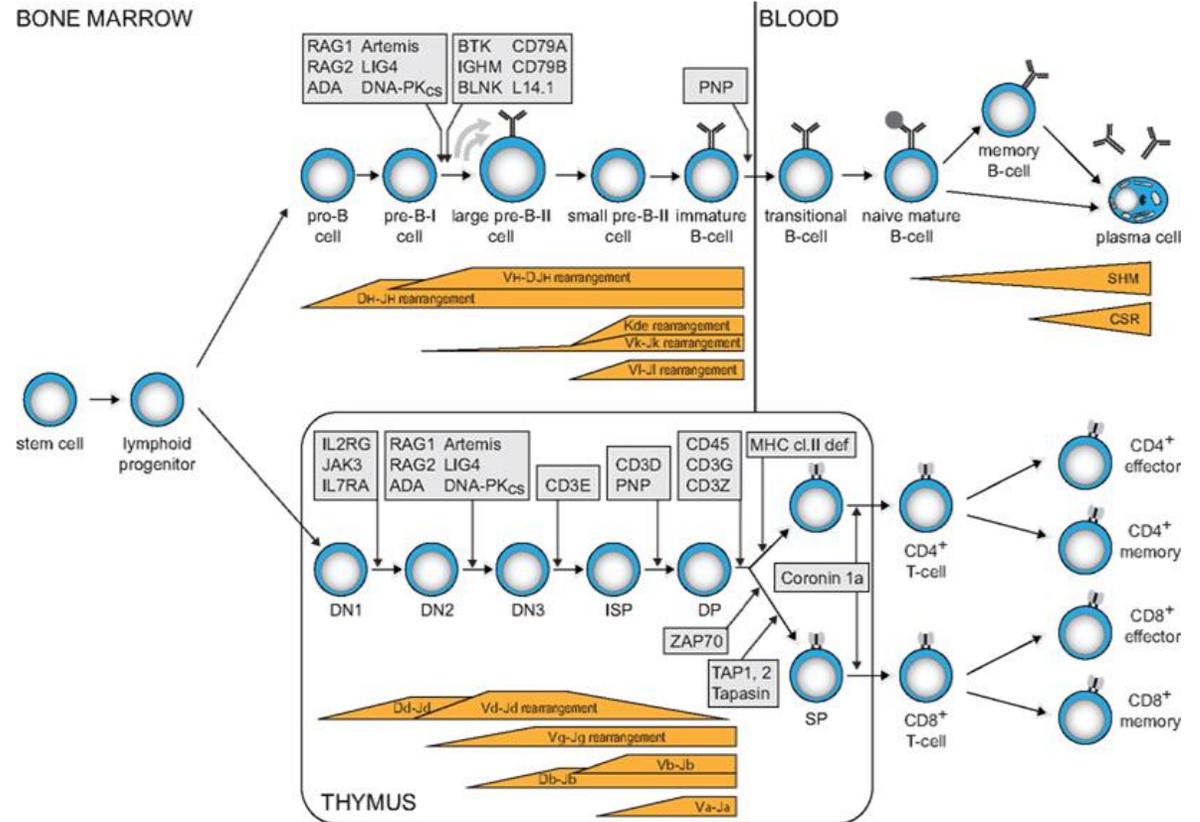


Imagem:
 Douglas Paes Barreto et al. *Jornal de Pediatria*: 2020.
 Online 22 de novembro.





Conteo hematológico completo
(números absolutos)

Inmunofenotipado por citometría de
flujo CD3, CD4, CD8, CD19, CD16+56
CD45RA+CD31+

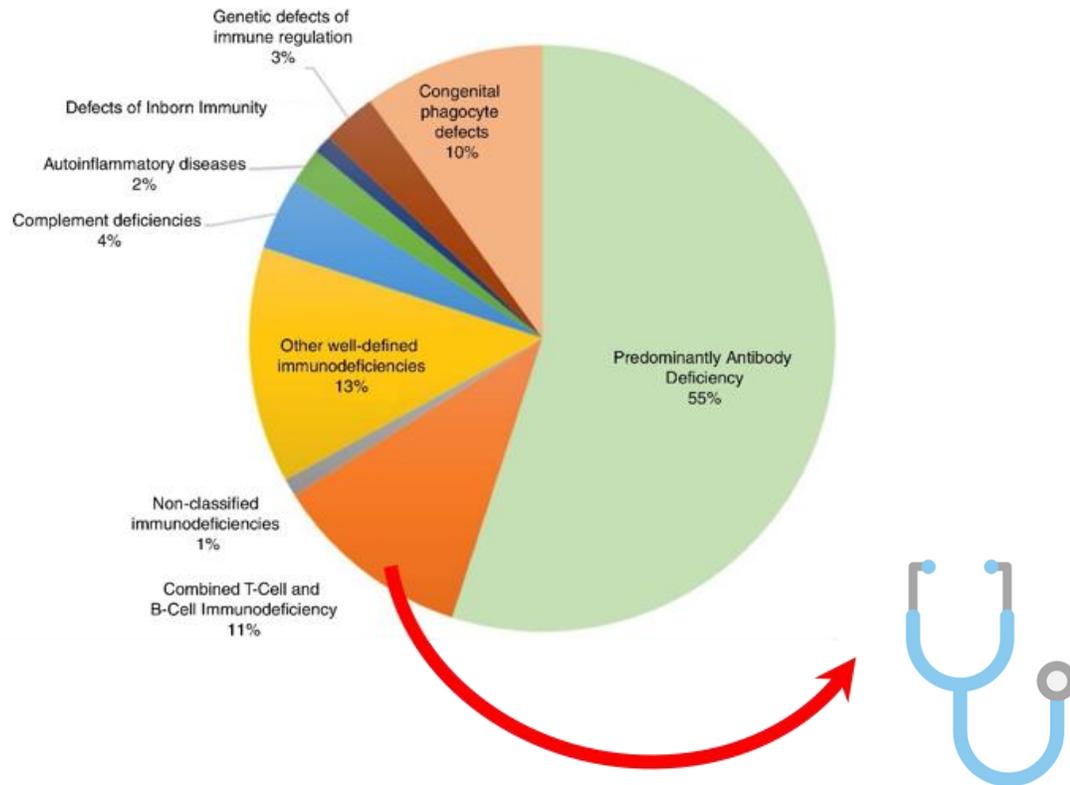
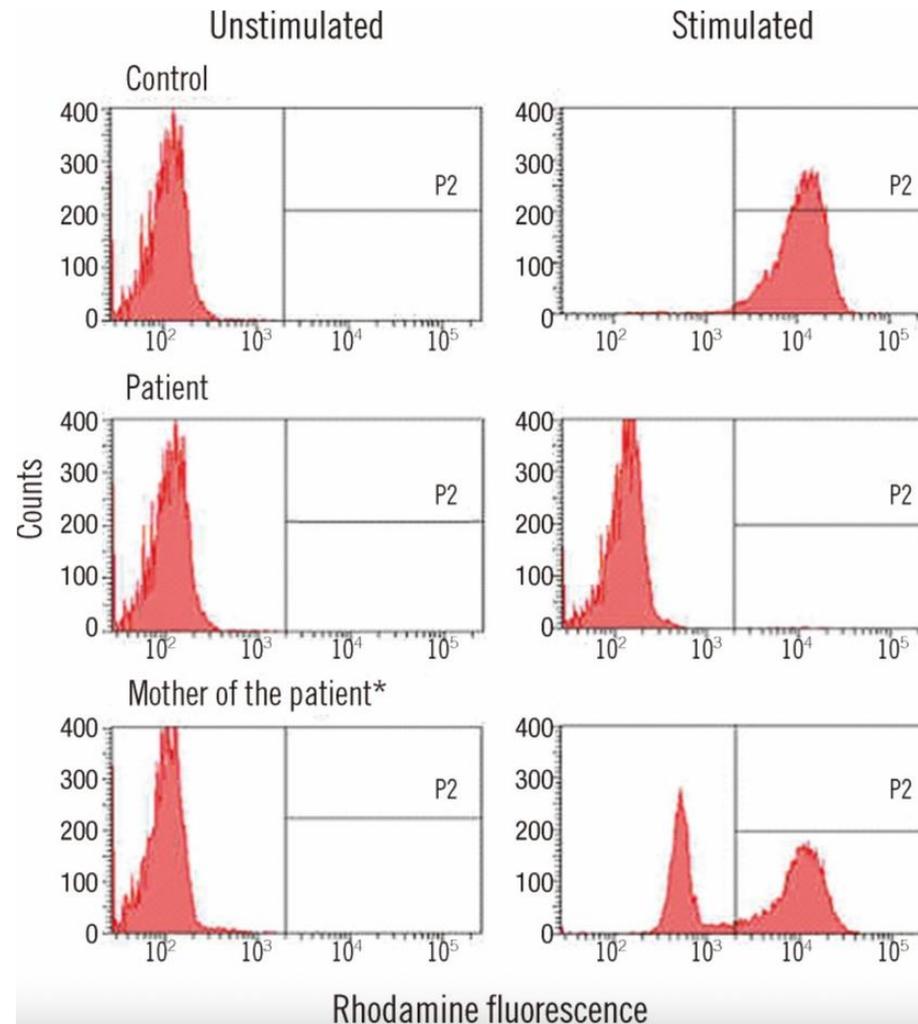
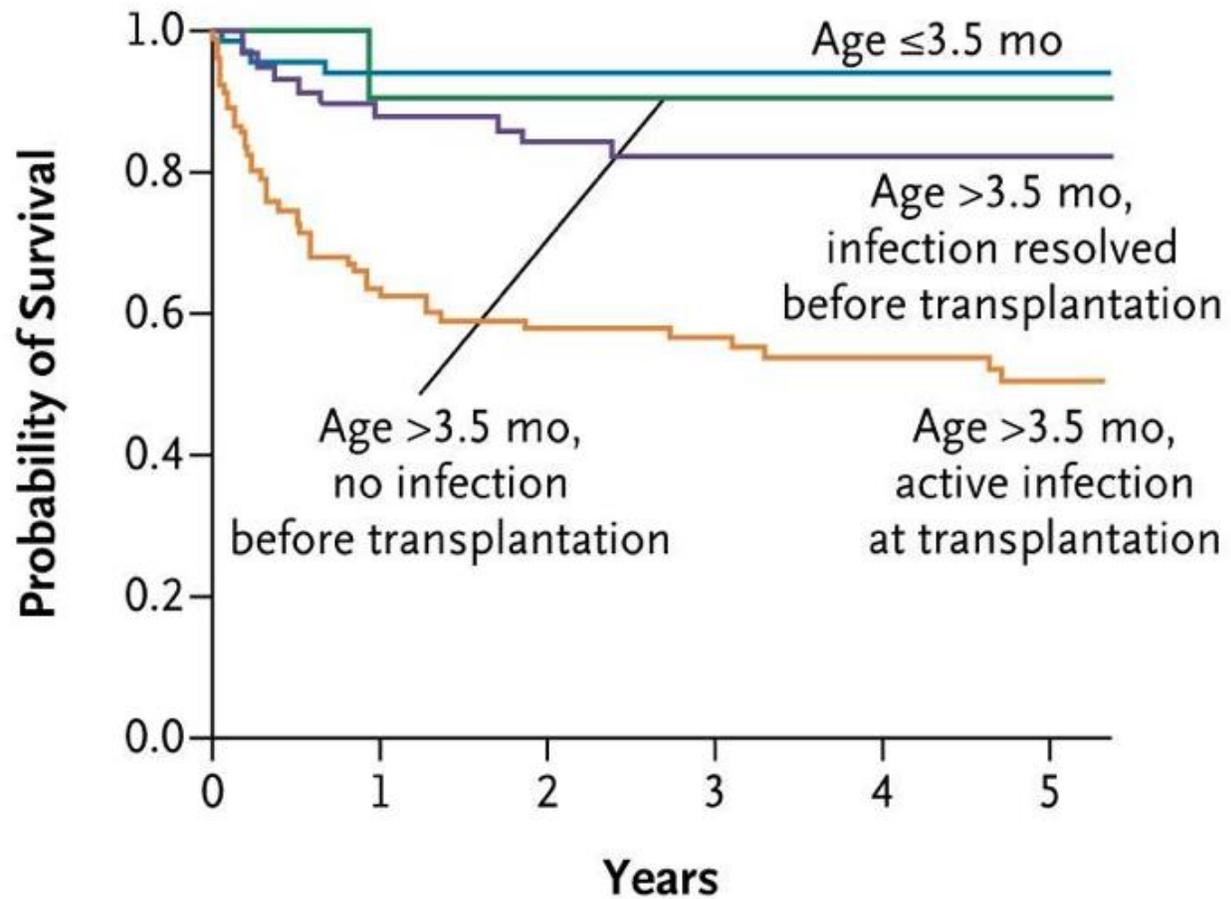


Imagem:
Douglas Paes Barreto et al. *Jornal de Pediatria*: 2020.
Online 22 de novembro.

Citometría de flujo
Prueba DHR
Enfermedad granulomatosa
crónica





240 bebés con IDCG
25 centros
2000-2009





TREC

Círculos de escisión del receptor de células T

KREC

Círculos de escisión de recombinación kappa



TREC

Círculos de escisión del receptor de células T

KREC

Círculos de escisión de recombinación kappa

¡CRIBADO!

Test de cribado neonatal

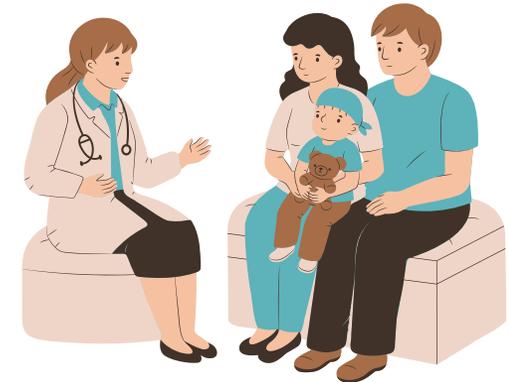


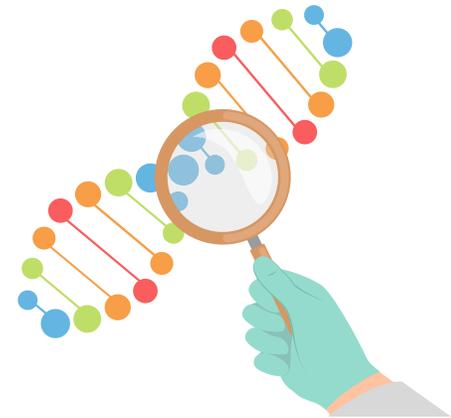
PROGRAMA de cribado neonatal

TEST de cribado neonatal



PROGRAMA de cribado neonatal



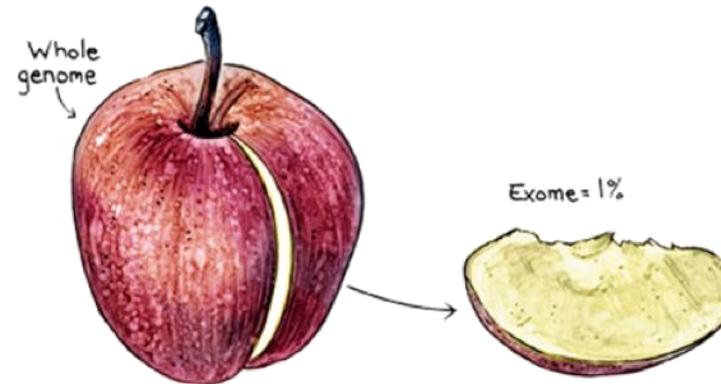
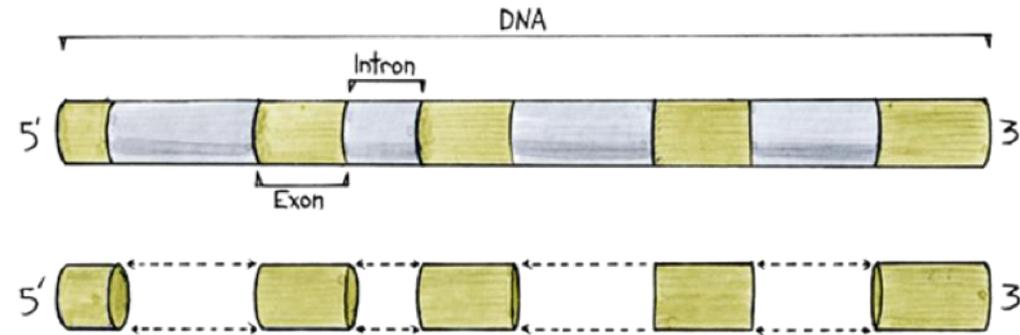


Sequenciación de próxima generación



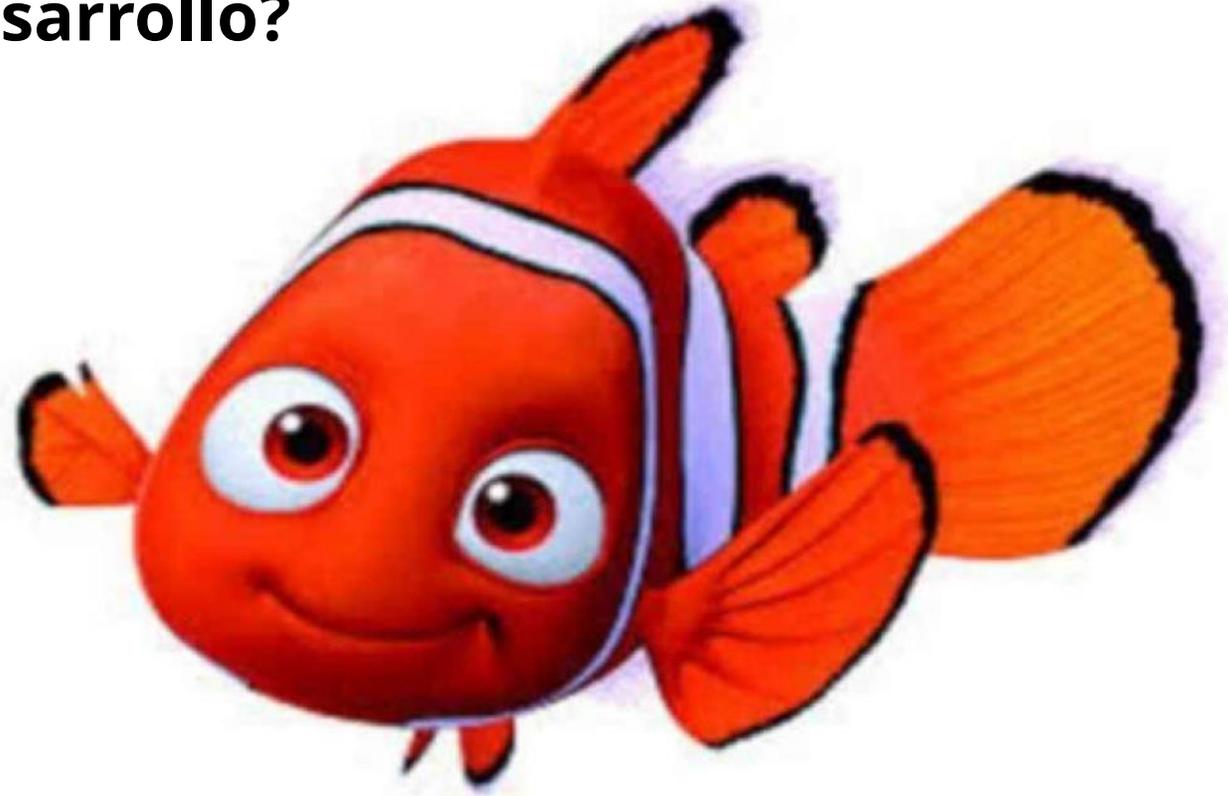
Sequenciación del genoma completo

Sequenciación del exoma completo



Copyright © 2012 University of Washington

¿Está el gen de su interés incluido en el panel o en desarrollo?



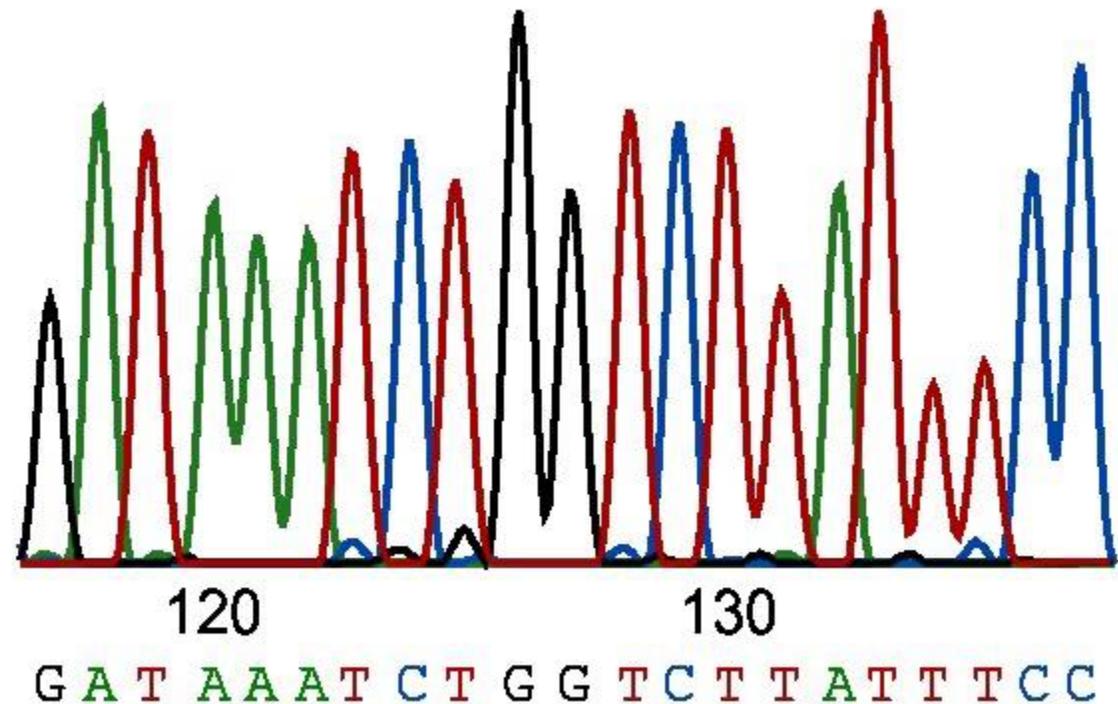
ACMG/AMP

		BENIGN CRITERIA		PATHOGENIC CRITERIA			
		Strong	Supporting	Supporting	Moderate	Strong	Very Strong
Strength of evidence		Strong	Supporting	Supporting	Moderate	Strong	Very Strong
Odds of Pathogenicity*		-18.7	-2.08	2.08	4.33	18.7	350.0
Evidence Category and Corresponding ACMG/AMP Codes	Population Data	BA1 ⁺ BS1 BS2			PM2	PS4	
	Allelic Evidence & Cosegregation Data	BS4	BP2 BP5	PP1			
					PM3 PM6	PS2	
	Computation & Predictive Data		BP1 BP3 BP4 BP7	PP2 PP3	PM1 PM4 PM5	PS1	PVS1
	Functional Data	BS3				PS3	
Other		BP6	PP4 PP5				

Patogénico
L. patogénico
VUS
 (variante de significado incierto)
L. benigno
Begnino

Sequenciación Sanger :

- Diagnóstico de los familiares
- confirmación de la variante identificada por secuenciación de próxima generación



› Zhonghua Er Ke Za Zhi. 2019 Dec 2;57(12):917-921. doi: 10.3760/cma.j.issn.0578-1310.2019.12.005.

[Application of copy number variation analysis based on raw data of next-generation sequencing in the molecular diagnosis for primary immunodeficiency disease]

[Article in Chinese]

Y Xia¹, X N Zhu, J Yang

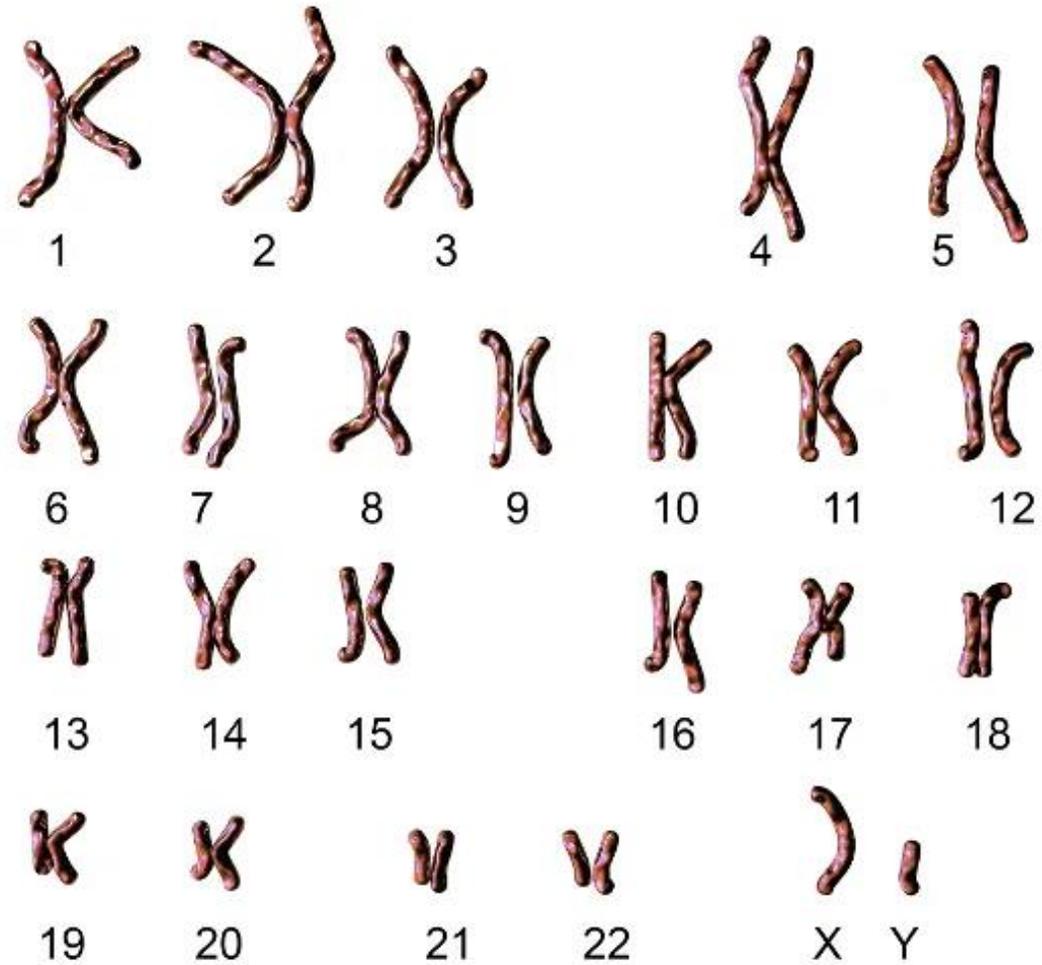
334 genes

Sequenciación de próxima generación – resultados negativos en 165 pacientes

Variación del número de copias en 95 (57,6%)

- 12 grandes eliminaciones en 12 pacientes
- Eliminación completa de un gen en 8 pacientes

CGH MLPA





Gracias!
carolina.prando@hpp.org.br

Q & A

COLLABORATION



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Avances en los tratamientos curativos y específicos

Advancements in curative and targeted treatments

Dr Pere Soler-Palacín, España
Dr Pere Soler-Palacín, Spain

Disclosures

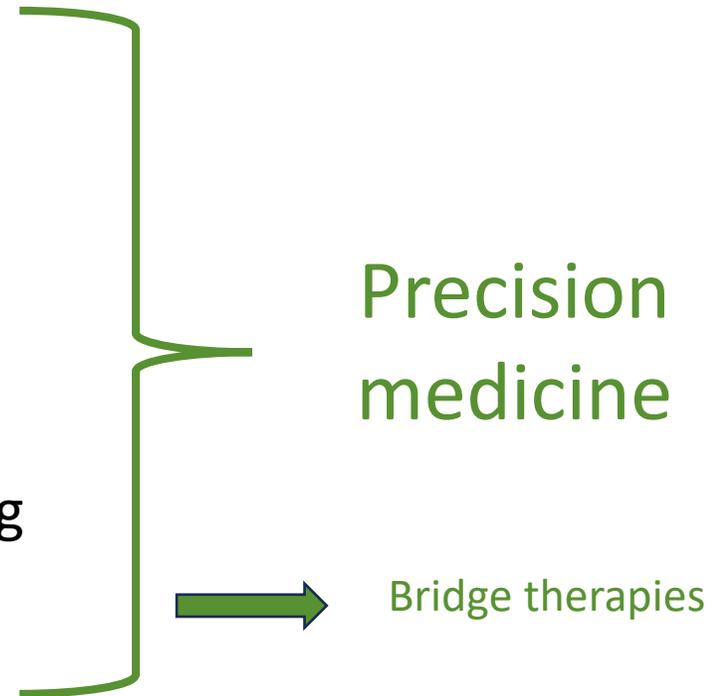
PSP has received grants from:

- ✓ CSL Behring
- ✓ Takeda
- ✓ Grifols
- ✓ Octapharma
- ✓ Binding Site
- ✓ UCB
- ✓ Pharming

But there are no conflict of interests regarding the content presented in this session

Therapeutic approaches in IEI

- Supportive vs **curative** treatments
 - Stem cell transplantation: new approaches
 - Gene therapy: new diseases
- **Targeted** vs unguided therapies
 - Biological therapies: the role of functional testing
 - Enzyme replacement therapy

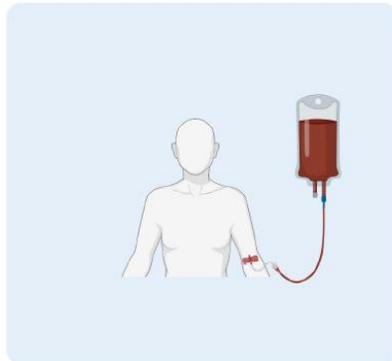


Precision
medicine

Bridge therapies

Therapeutic approaches in IEI

HSCT



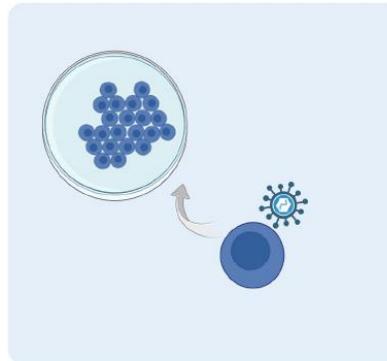
Pros:

- Curative
- Well known technique
- Experience in different conditioning regimes

Cons:

- GVHD
- Risk of adverse events (like infection) during conditioning or after transplant

Gene therapy



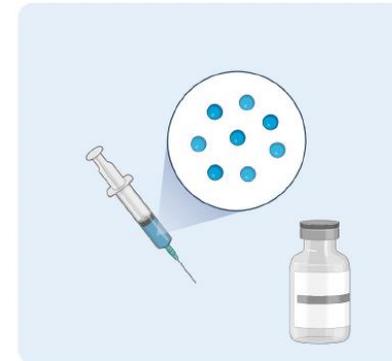
Pros:

- Potentially curative
- Not limited by donor availability
- Eliminates GVHD
- Reduced risk associated to conditioning regimes

Cons:

- Currently still not applicable to the majority of PID
- Lack or low availability
- Long term side effects not fully established

Biologic or small molecules



Pros:

- Target only the molecules involved
- Quicker control of disease symptoms
- Easier access in a higher number of centers
- Transitioning treatment for HSCT

Cons:

- No definitive cure
- Long term side effects not fully established
- Risk of adverse events (like infection) during treatment

Therapeutic approaches in IEI

- Supportive vs **curative** treatments
 - Stem cell transplantation: new approaches
 - Gene therapy: new diseases
- **Targeted** vs unguided therapies
 - Biological therapies: the role of functional testing
 - Enzyme replacement therapy

Recent advances in curative therapies

Illustrations: Lienke Post

Disadvantages of current treatment



Lack of suitable donors.



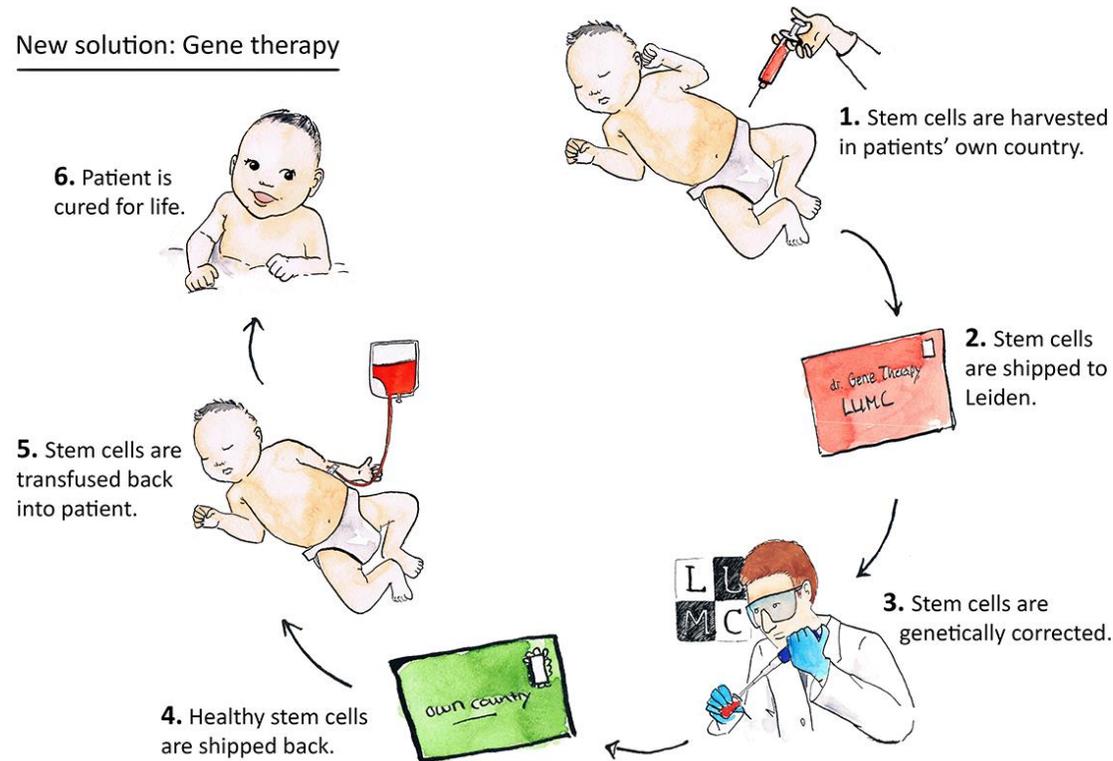
Chance of severe adverse effects.



Patient and family have to travel.

Recent advances in curative therapies

New solution: Gene therapy

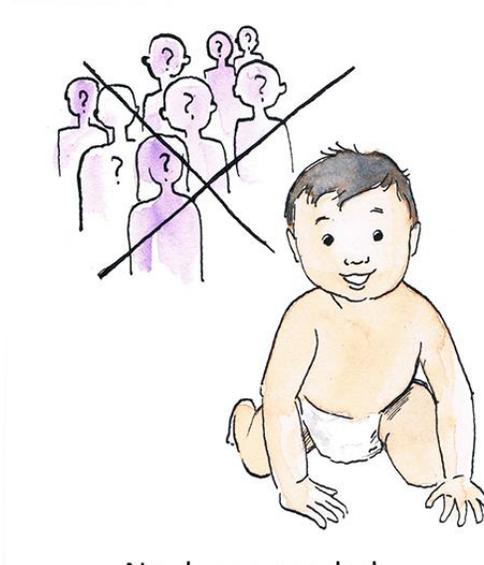


Illustrates: Lieneke Post

Recent advances in curative therapies

Illustrations: Lieneke Post

Advantages of Gene Therapy



No donor needed.

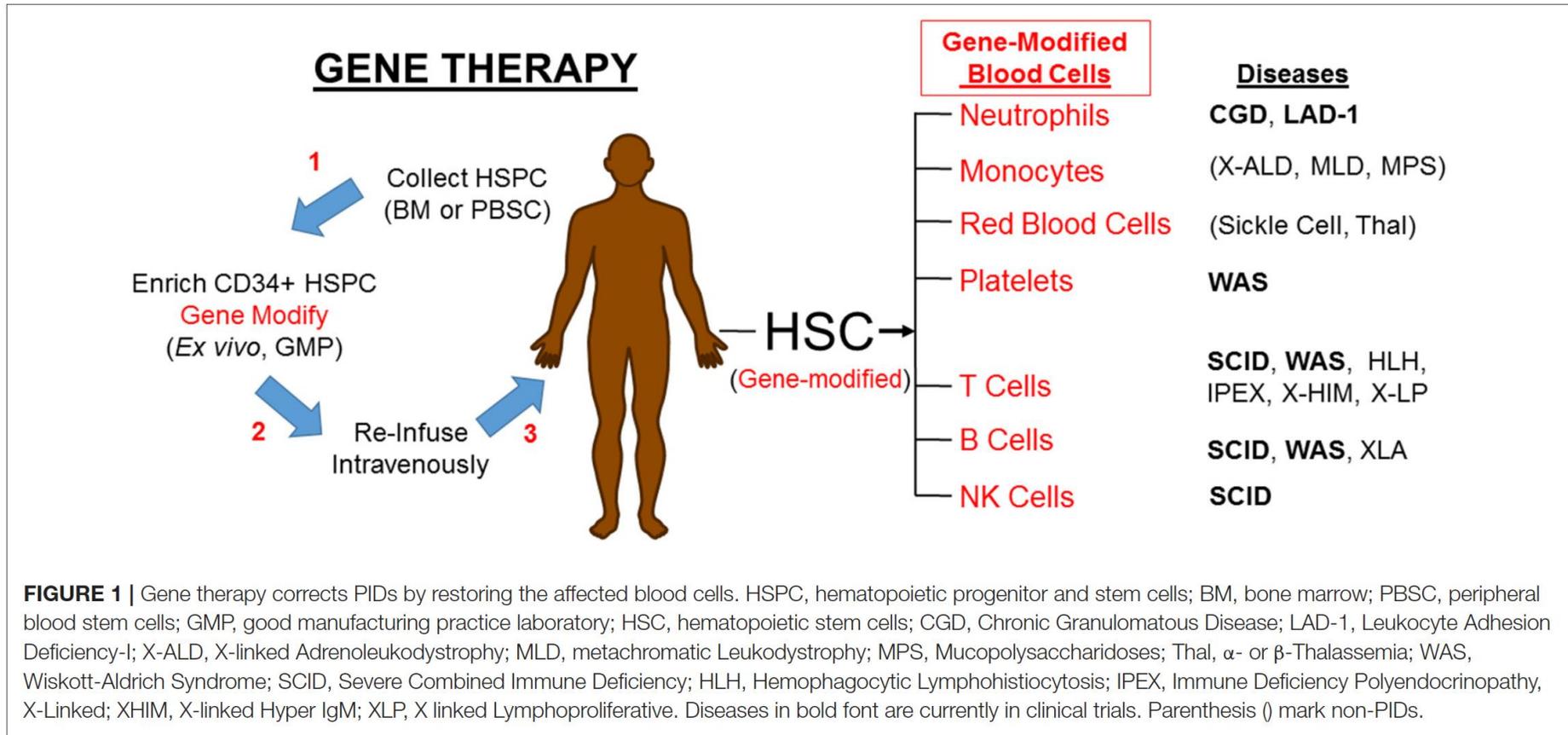


No use of foreign material,
making it safer.



Treatment is travelling,
family stays home.

Gene therapy for IEI patients



Gene therapy for SCID patients: it all started in 2000

Gene Therapy of Human Severe Combined Immunodeficiency (SCID)-X1 Disease

Marina Cavazzana-Calvo,^{1,2,3} Salima Hacein-Bey,^{1,2,3}
Geneviève de Saint Basile,¹ Fabian Gross,² Eric Yvon,³
Patrick Nusbaum,² Françoise Selz,¹ Christophe Hue,^{1,2}
Stéphanie Certain,¹ Jean-Laurent Casanova,^{1,4} Philippe Bousso,⁵
Françoise Le Deist,¹ Alain Fischer^{1,2,4†}



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NUMBER 16



SUSTAINED CORRECTION OF X-LINKED SEVERE COMBINED IMMUNODEFICIENCY BY EX VIVO GENE THERAPY

SALIMA HACEIN-BEY-ABINA, PH.D., FRANÇOISE LE DEIST, M.D., PH.D., FRÉDÉRIQUE CARLIER, B.S., CÉCILE BOUNEAUD, PH.D., CHRISTOPHE HUE, B.S., JEAN-PIERRE DE VILLARTAY, PH.D., ADRIAN J. THRASHER, M.D., PH.D., NICOLAS WULFFRAAT, M.D., RICARDO SORENSEN, M.D., SOPHIE DUPUIS-GIROD, M.D., ALAIN FISCHER, M.D., PH.D., AND MARINA CAVAZZANA-CALVO, M.D., PH.D.

Correction of ADA-SCID by Stem Cell Gene Therapy Combined with Nonmyeloablative Conditioning

Alessandro Aiuti,¹ Shimon Slavin,² Memet Aker,²
Francesca Ficara,¹ Sara Deola,¹ Alessandra Mortellaro,¹
Shoshana Morecki,² Grazia Andolfi,¹ Antonella Tabucchi,³
Filippo Carlucci,³ Enrico Marinello,³ Federica Cattaneo,¹
Sergio Vai,¹ Paolo Servida,⁴ Roberto Miniero,⁵
Maria Grazia Roncarolo,^{1,6} * Claudio Bordignon^{1,6*†}



The NEW ENGLAND JOURNAL of MEDICINE

ESTABLISHED IN 1812

JANUARY 29, 2009

VOL. 360 NO. 5

Gene Therapy for Immunodeficiency Due to Adenosine Deaminase Deficiency

Alessandro Aiuti, M.D., Ph.D., Federica Cattaneo, M.D., Stefania Galimberti, Ph.D., Ulrike Benninghoff, M.D., Barbara Cassani, Ph.D., Luciano Callegaro, R.N., Samantha Scaramuzza, Ph.D., Grazia Andolfi, Massimiliano Mirolo, B.Sc., Immacolata Brigida, B.Sc., Antonella Tabucchi, Ph.D., Filippo Carlucci, Ph.D., Martha Eibl, M.D., Memet Aker, M.D., Shimon Slavin, M.D., Hamoud Al-Mousa, M.D., Abdulaziz Al Ghonaim, M.D., Alina Ferster, M.D., Andrea Duppenhaler, M.D., Luigi Notarangelo, M.D., Uwe Wintergerst, M.D., Rebecca H. Buckley, M.D., Marco Bregni, M.D., Sarah Markt, M.D., Maria Grazia Valsecchi, Ph.D., Paolo Rossi, M.D., Fabio Ciceri, M.D., Roberto Miniero, M.D., Claudio Bordignon, M.D., and Maria-Grazia Roncarolo, M.D.

Gene therapy for RAG1-SCID patients



Partners

The Netherlands

LUMC (Leiden)
BBS (Leiden)

France

INSERM (Paris)
SCETIDE (Paris)

Germany

Genewerk (Heidelberg)
LMU (Munich)
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UULM (ULM)

Israel

SHEBA (Tel Aviv)

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OPBG (Rome)
OSR (Milan)

Poland

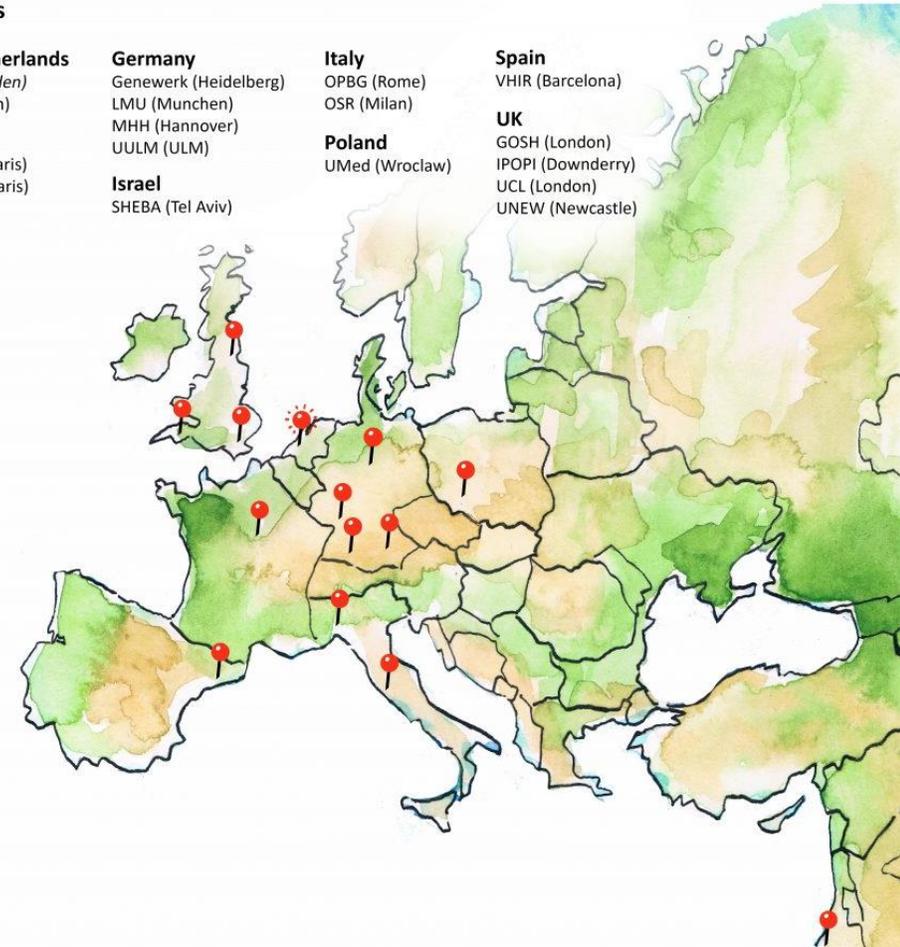
UMed (Wroclaw)

Spain

VHIR (Barcelona)

UK

GOSH (London)
IPOPI (Downerry)
UCL (London)
UNEW (Newcastle)



Gene therapy for Artemis patients

THE NEW ENGLAND JOURNAL OF MEDICINE

RESEARCH SUMMARY

Lentiviral Gene Therapy for Artemis-Deficient SCID

Cowan MJ et al. DOI: 10.1056/NEJMoa2206575

CLINICAL PROBLEM

Artemis-deficient severe combined immunodeficiency (ART-SCID), resulting from damaging variants in the gene *DCLRE1C*, accounts for 2 to 3% of all SCID cases. ART-SCID responds poorly to allogeneic hematopoietic-cell transplantation, which underscores the need for alternative treatments.

CLINICAL STUDY

Design: A phase 1-2, single-center, nonrandomized clinical study evaluated the effects of transfusion of autologous CD34+ bone marrow cells, transfected with a lentiviral vector containing *DCLRE1C* complementary DNA and its natural promoter, in infants with newly diagnosed ART-SCID.

Intervention: 10 infants first underwent bone marrow harvest for production of lentiviral *DCLRE1C*-corrected CD34+ cells. They then received conditioning with intravenous low-dose busulfan over a period of 2 days, followed 1 day later by infusion of the CD34+ cells. End points included safety and T-cell reconstitution.

RESULTS

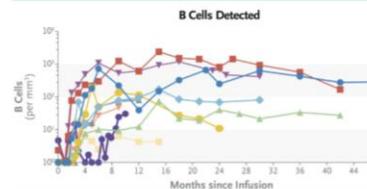
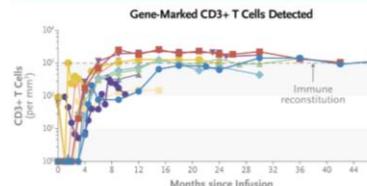
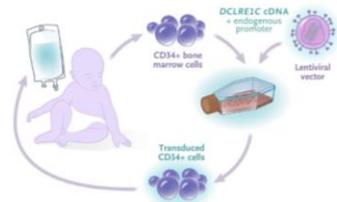
Safety: Busulfan toxicity manifested as transient blood cytopenias, of which 16 were grade 3 or 4. Autoimmune hemolytic anemia developed in four patients 4 to 11 months after infusion; all cases resolved with immune reconstitution.

Immune Reconstitution: Gene-marked CD3+ T cells were detected at a median of 12 weeks in all 10 patients. Of 9 patients followed for at least 12 months, 4 had T-cell immune reconstitution at the 12-month mark. Of 6 patients followed for at least 24 months, 5 had T-cell reconstitution at a median of 12 months (range, 6 to 24 months). B cells were detected by flow cytometry and gene marking in all 10 patients, 4 of whom were able to stop immune globulin infusions.

LIMITATIONS AND REMAINING QUESTIONS

- Larger studies of longer duration are needed to further assess the safety and efficacy of this approach.

Links: Full Article | NEJM Quick Take | Science behind the Study



CONCLUSIONS

Among infants with newly diagnosed Artemis-deficient severe combined immunodeficiency, infusion of autologous lentiviral gene-corrected CD34+ bone marrow cells after conditioning with low-dose busulfan resulted in gene-corrected, functional T cells and B cells and the expected grade 3 or 4 adverse events with chemotherapy.

Especially important due to limited results with SCT and radiosensitivity

Stem cell transplant: “modified” haplo SCT



1. Donor availability for inborn errors of immunity (IEI), before 2010 versus after 2010

Importance of optimizing transplant strategy for mismatched/haploidentical donor transplant

2. Improved haplo HSCT outcomes using TCR $\alpha\beta$ /CD19 depletion and post-cyclophosphamide (PTCY)

3. Comparison between TCR $\alpha\beta$ /CD19 depletion and PTCY strategies

Real world data from EBMT IEWP study – 40 participating centres

4. Haploidentical versus matched family/unrelated donor transplant (MFD/MUD)

PTCY versus MFD/MUD – no published data

TCR $\alpha\beta$ /CD19 depletion versus MFD/MUD – published data

Major challenges for developing universal donors and a potential solution using memory T cell addback

Donor availability should no longer be a problem



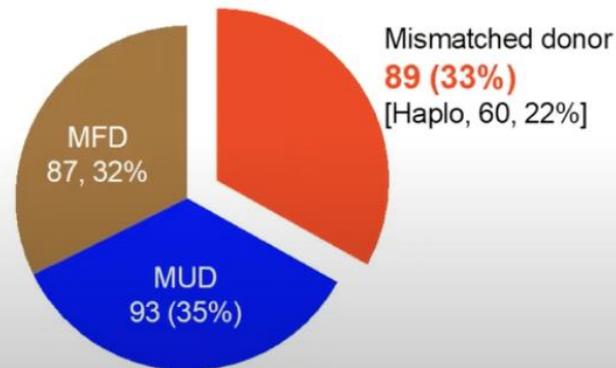
Stem cell transplant: “modified” haplo SCT



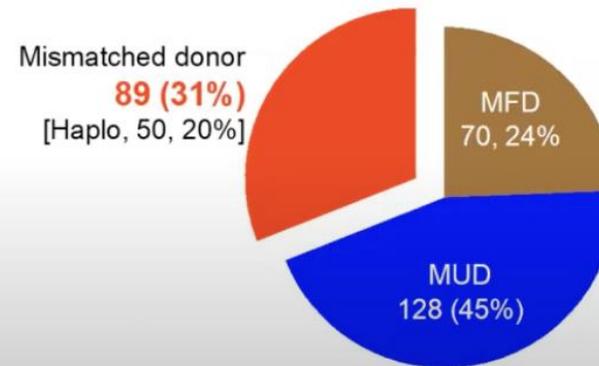
Donor availability for children with IEI in Newcastle upon Tyne, UK

A third of patients do not have a suitably matched donor

First transplant from 1987 – 2009
(n=269)



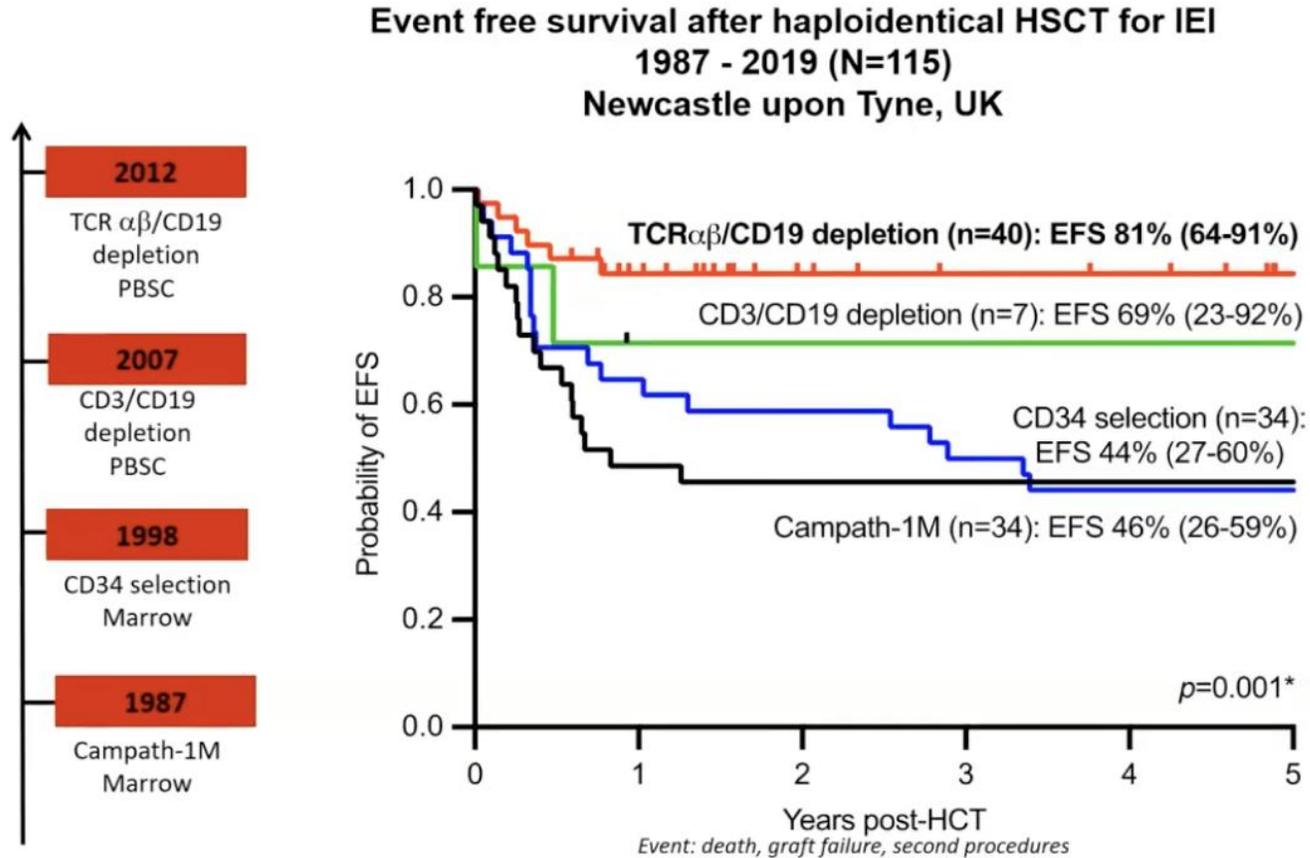
First transplant from 2010-2019
(n=287)



Unpublished data



Stem cell transplant: “modified” haplo SCT



Lum et al, BMT 2020 Updated

Stem cell transplant: "modified" haplo SCT

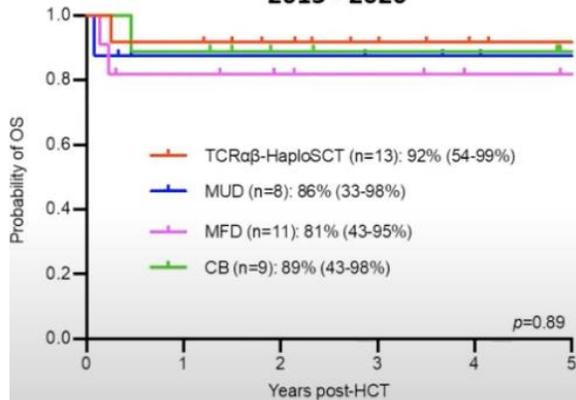


TCRαβ-HaploSCT versus matched family/unrelated donor transplant Newcastle upon Tyne

All received Treosulfan based conditioning since 2007
Alemtuzumab in MFD/MUD; ATG in TCRαβ-HaploSCT

Severe combined immunodeficiency (SCID)

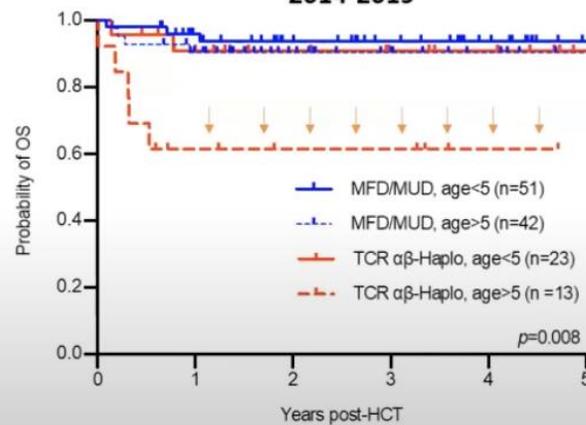
N=41
2013 - 2020



Christo et al J Clin Immunology 2022

Non-SCID IEI

N=129
2014-2019



Lum et al Blood advances 2022

Grade II-IV aGvHD
TCRαβ-HaploSCT: 18%
MUD PBSC: 26%
MFD marrow: 18%

None had cGvHD in the entire cohort

3 major obstacles for developing universal donors

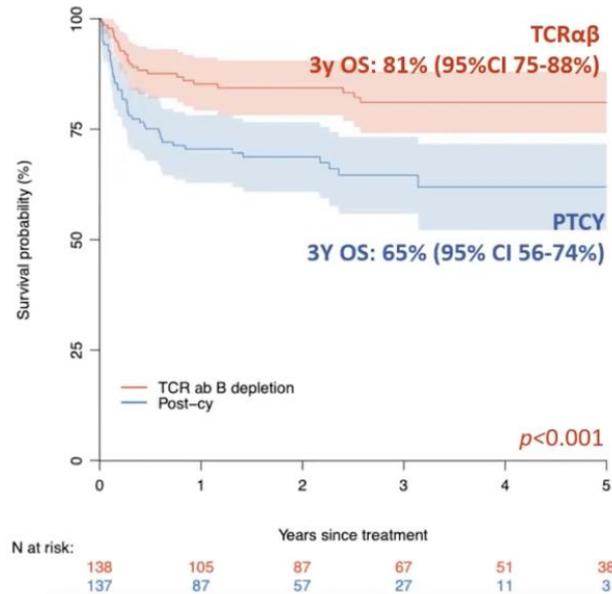
1. Delayed immune reconstitution
2. Increased viraemia
3. Excess transplant related mortality in older patients (due to infection)



Stem cell transplant: “modified” haplo SCT



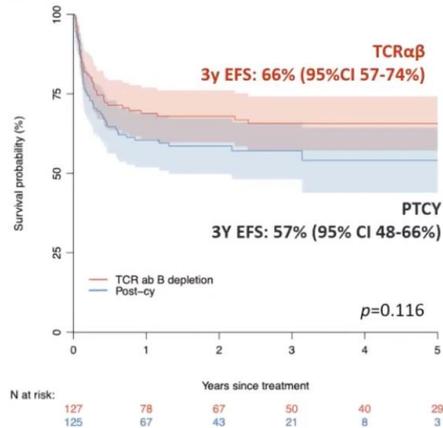
Overall survival was significantly higher after TCR $\alpha\beta$



Stem cell transplant: “modified” haplo SCT

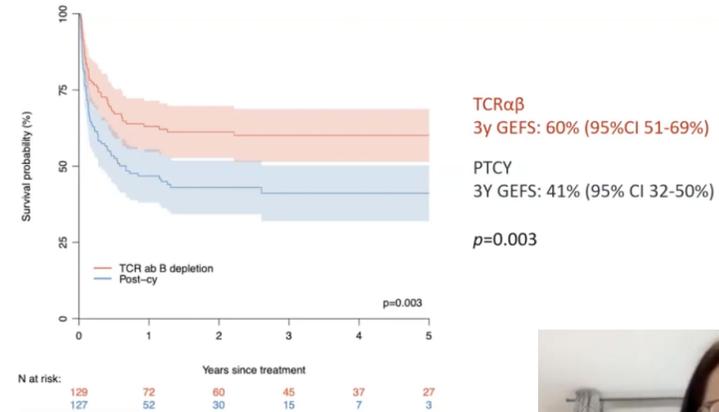


Event-free survival
 Event free survival is defined as survival without graft failure and second procedures
No significant difference between TCRαβ and PTCY



GvHD-free, event-free survival (GEFS)
 GEFS is defined as survival without graft failure, second procedures/transplant, grade III-IV aGvHD and chronic GvHD

GEFS was significantly higher after TCRαβ



Stem cell transplant: also for adult ICI patients

YIN AND YANG OF AUTOIMMUNITY AND IMMUNODEFICIENCIES IN HEMATOLOGY



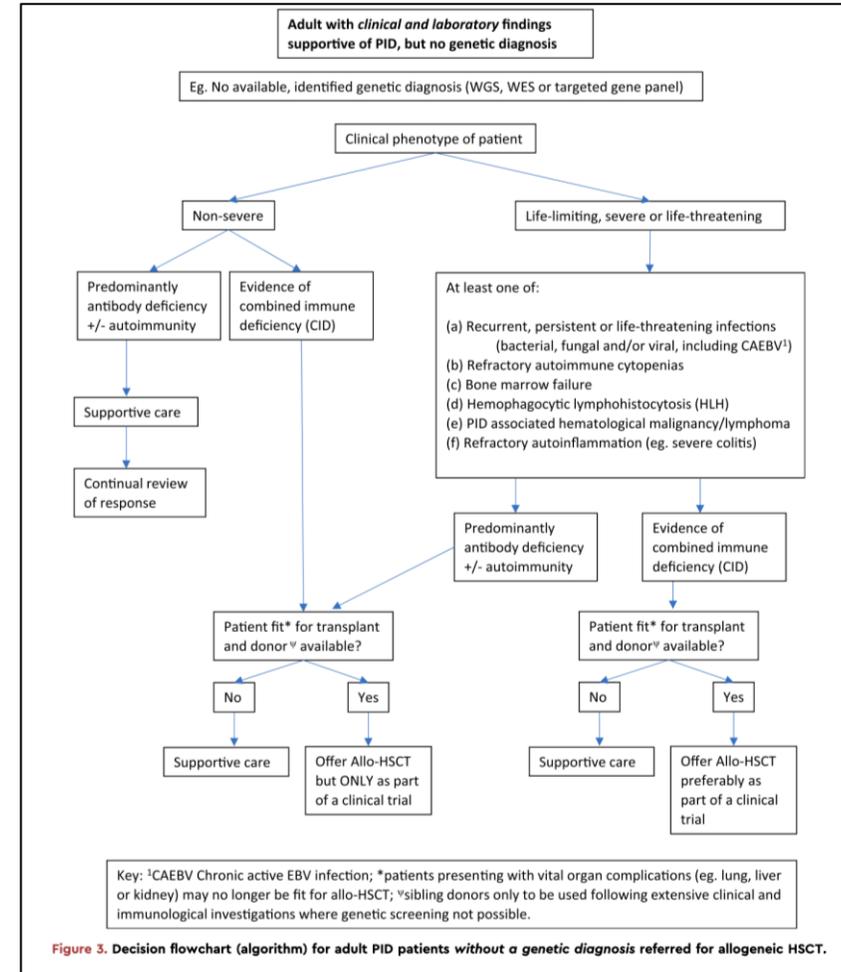
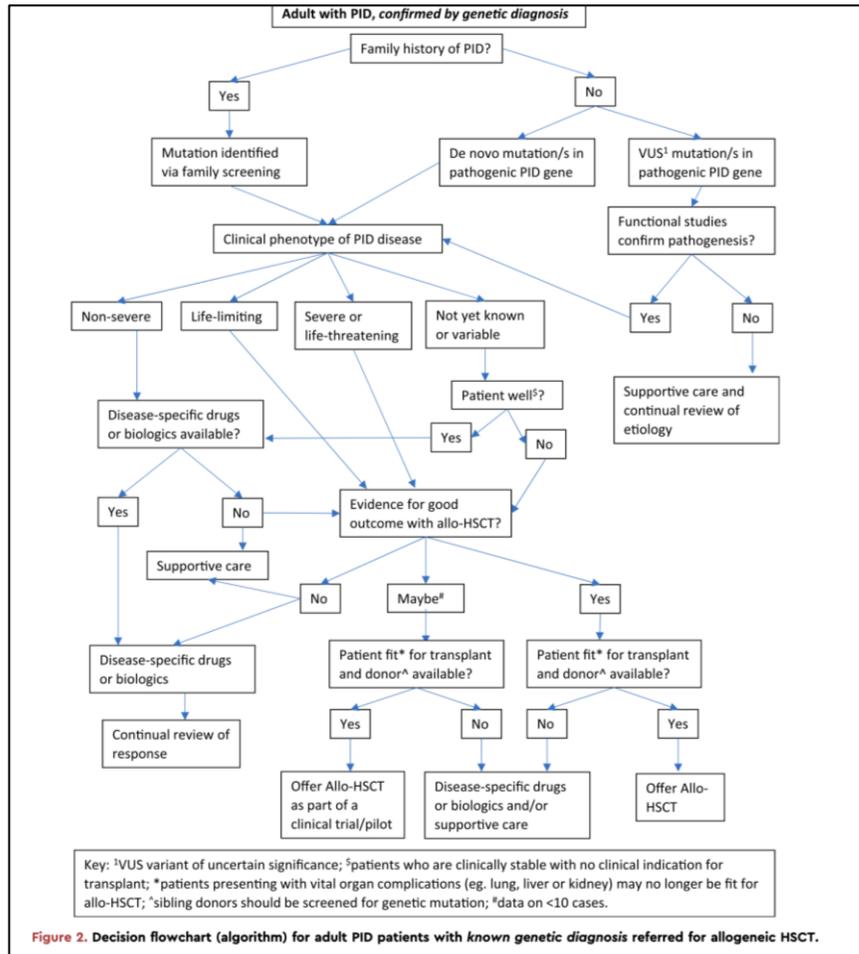
Allogeneic hematopoietic stem cell transplantation in adults with primary immunodeficiency

Emma C. Morris
Institute of Immunity and Transplantation, University College London, London, United Kingdom

Table 1. Summary of published allo-HSCT outcome data for adult (and adolescent) PID patients

Reference	No. of patients	PID subtype	Age at HSCT, y (range)	Donor (no.)	Conditioning	OS	EFS	TRM	Engraftment	Median follow-up (range)
Albert et al 2018 ⁴	18	6 CGD 12 other PID	18 y (15-22)	MRD (2) MUD (5) 1Ag MMUD (1)	Full Bu/Cy (2); Full Bu/Flu (1); Sub Bu/Flu (7); Flu/Mel ±TT (1); Treo/Flu ±TT (7). All had serotherapy.	94%	94%	6%	100%	5 y (2-9 y)
Fox et al 2018 ⁵	29	11 CGD 18 other PID	24 y (17-50)	MRD (1) MUD (13) 1Ag MMUD (5)	Flu/Mel/Alem (20) Flu/Bu/ATG (8) Flu/Bu/Alem (1)	89% at 1yr 85% at 3yrs	90%	14%	100%	3.5 y (4 mo to 12 y)
Jin et al 2018 ¹⁸	8	Primary HLH	25 y (18-54)*	HaploID (6) MUD (2)	TBI/VP16/Cyclo (6); VP16/Flu/Bu/ATG (2)	88%	NS	12%	100%	27 mo
Leiding et al 2018 ¹⁹	5 (AYA) 10 (ped <12 y)	STAT1 GOF	29 y (18-35) 8 y (1-17)	MRD (4); MUD (8); MMUD (1); HaploID (2)*; UCB (2, 2)*.	Flu/Mel/Alem (4); Bu/Cy (3); Flu/Bu or Treo/ATG or Alem (6); Various other (4*)	20% at 1yr (AYA) 60% at 1yr (ped) 40% at 1yr (all)	NS NS NS	NS	50%*	NS
Parta et al 2017 ²⁰	17 (AYA) 20 (ped)	CGD	24 y (18-32) 8 y (4-17)	MRD (6); MUD (30); MMUD (1)	Bu/Alem (6); Bu/TBI/Alem (3)	82.5% (all)	80% (all)	17.5% (all)	85% (all)	3.4 y (range, NS)
Shah et al 2017 ²¹	7	DOCK8 Defic	20 y (7-25)	HaploID (7)	Flu/Bu/Cy + low dose TBI (7)	86% (all)	NS	14%	100%	2 y (3 mo to 5.7 y)
Fu et al 2016 ²²	30	1° HLH; EBV-HLH; Tu-HLH; Undef- HLH	23 y (14-52) 19 y (14-55) 24 y (14-44) 29 y (16-32)	HaploID (23); MRD (6); MUD (1).	Bu/Cyclo/VP16 (6) for MRD TBI/Cyclo/VP16 + ATG (24) for HaploID/MUD	63.3% at 2 y (100% in 1° HLH; 64% in EBV-HLH; 17.7% in Tu-HLH; 25% in Undef)	NS		100%	26 mo
Wehr et al 2015 ¹¹	14 (adult) 11 (ped)	CVID	34 y (18-50) 14 y (8-17)	MRD (14); MUD (10); MMUD (1)	BCNU/Flu/Mel (5); Flu/Mel (7); Flu/Mel/Treo (2); Flu/Bu ±TT (4); Bu/Cy ±AraC (6); Flu/Cy (1).	57% (adults) 52% (all patients)	14%	44% at 1 y (all)	79% (adult)	NS
Grossman et al 2014 ²³	14	GATA2 Defic	33 y (15-46)	MRD (4); MUD (4); UCB (4); HaploID (2).	Flu/low dose TBI (8); Flu/Cy/low dose TBI (6).	57% (all)	NS	28%	100%	3.5 y (1-5 y)
Gungor et al 2014 ²⁴	13 (adult) 43 (ped)	CGD CGD	21 y (18-39) 9 y (0.8 - 17)	MRD (21); MUD (25); MMUD (10).	Flu/Bu ATG or Alem (all).	92% (adult) 96% (all)	91% (all)	7%	93% (adult)	21 mo
Spinner et al 2017 ²⁵	21	GATA2 Defic	NS (15-49 y)	NS	NS	72% at 1 y; 65% at 2 y; 54% at 4 y	NS	NS	NS	14 mo (0-180 mo)

Stem cell transplant: also for adult IEL patients



Therapeutic approaches in IEI

- Supportive vs **curative** treatments
 - Stem cell transplantation: new approaches
 - Gene therapy: new diseases
- **Targeted** vs unguided therapies
 - Biological therapies: the role of functional testing
 - Enzyme replacement therapy

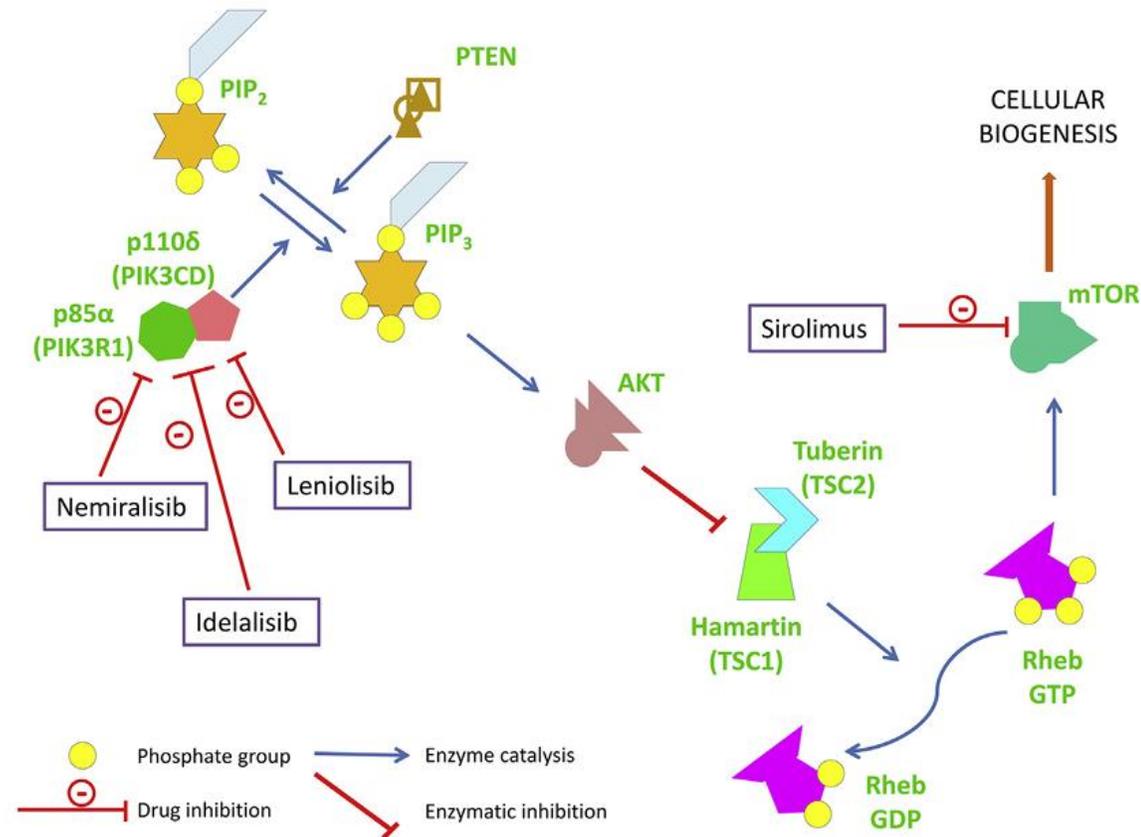
Targeted therapies

Targeted therapies specifically target molecular components of the immune system. In doing so these targeted therapies inhibit, enhance or restore the balance of the specific component(s) of the immune system that are not functioning properly in a given PID.



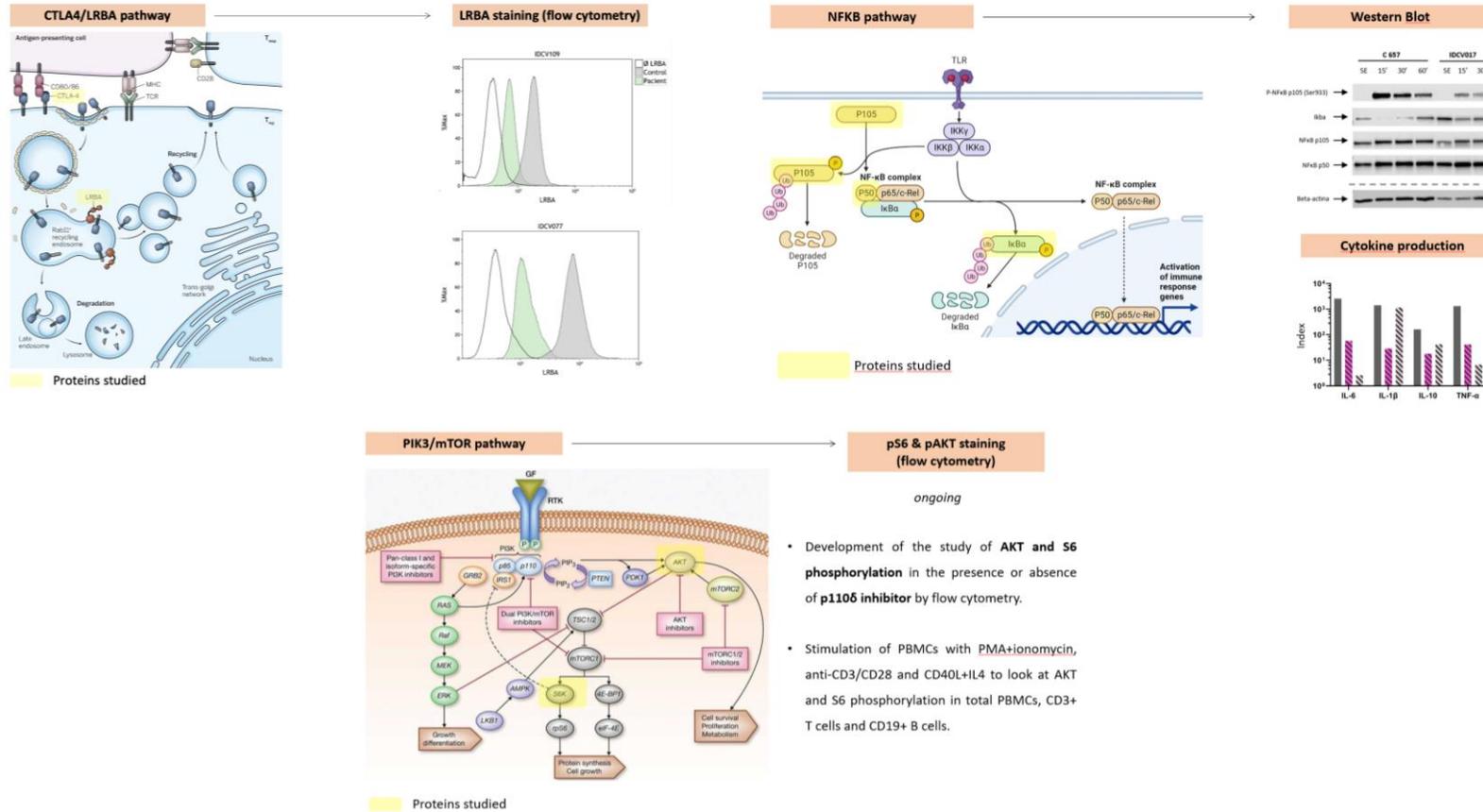
Targeted therapies

Personalized treatment can be used when we know the molecular mechanisms underlying the disease



Targeted therapies

Even without knowing molecular underlying mechanisms, functional testing may allow this approach



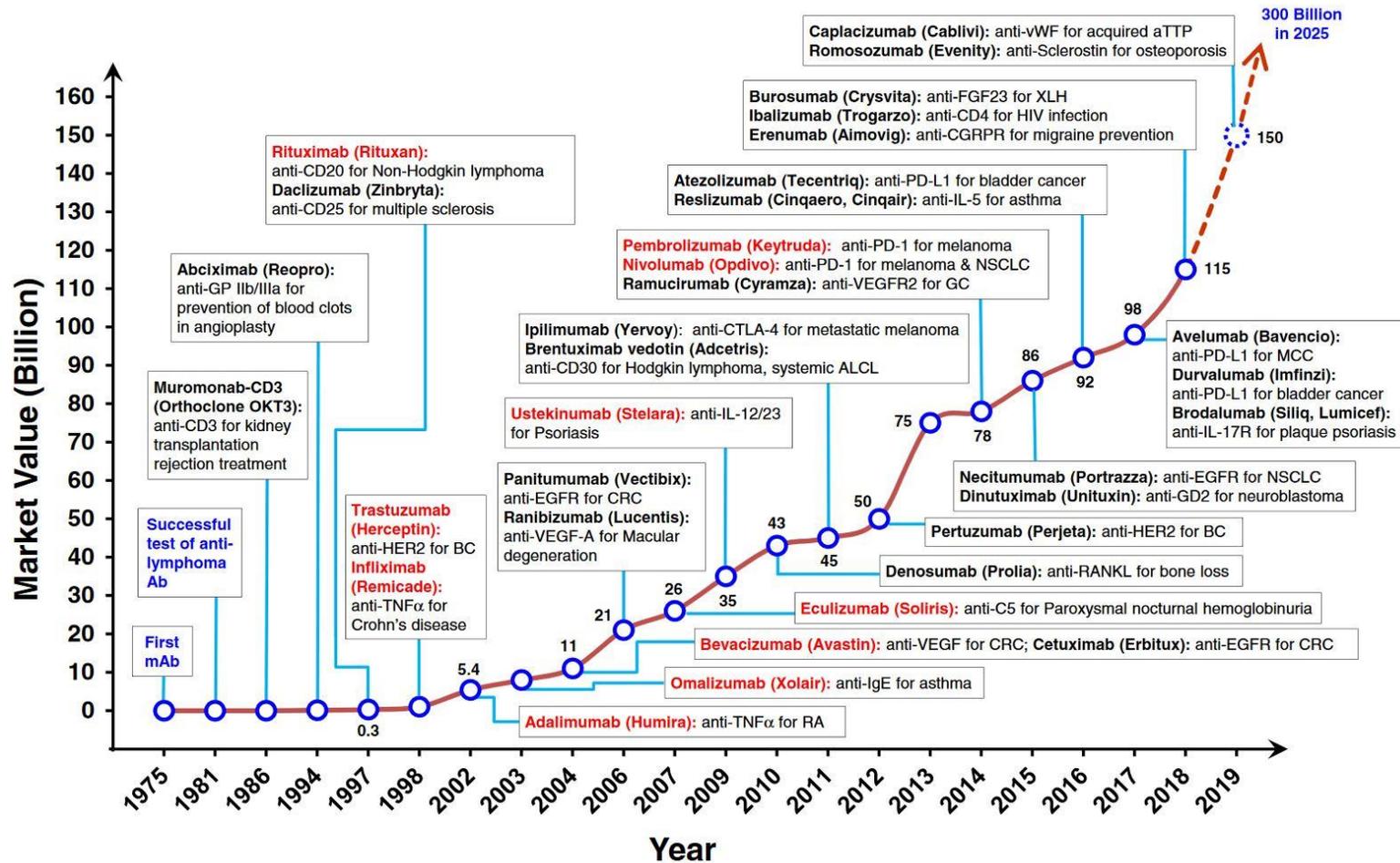
Targeted therapies: biologicals and small molecules

TARGETED THERAPIES AND THEIR MOLECULAR TARGETS ¹			
Targeted therapy	Molecular structure	Molecular target	Potential PID indications
Biological agents			
Abatacept Belatacept	CTLA-4 IgG fusion protein	B7-1 (CD80), B7-2 (CD86)	CTLA-4 haploinsufficiency, LRBA deficiency
Anakinra	Recombinant human IL-1R antagonist	IL-1R	Cryopyrin-associated periodic fever syndromes
Canakinumab	Antihuman IL-1 IgG1 mAb	IL-1β	CAPS, FCAS
Rilonacept	IgG1 linked to IL-1R and IL-1R accessory protein		MWS, DIRA
Etanercept	Fusion protein	TNF-α	SAV1
Infliximab	Chimeric mAb		CANDLE syndrome
Adalimumab	Humanized mAb		POMP deficiency
Tocilizumab	IgG1k recombinant humanized mAb	IL-6R	STAT3-GOF

TARGETED THERAPIES AND THEIR MOLECULAR TARGETS ¹			
Small molecule drugs			
Sirolimus	Macrolide compound	mTOR	NLCR4-GOF POMP deficiency CTLA-4 haploinsufficiency APDS
Ruxolitinib Baricitinib	Pyrazole molecules	JAK1, JAK2, JAK3	STAT3-GOF* STAT1-GOF CANDLE syndrome APDS
Tofacitinib	Pyrrolopyrimidine		
Leniolisib	Pyrrolopyrimidine	PI3K-delta inhibitor	APDS
Tadekinig-α	Recombinant IL-18 binding protein	IL-18 binding protein	NLCR4-GOF

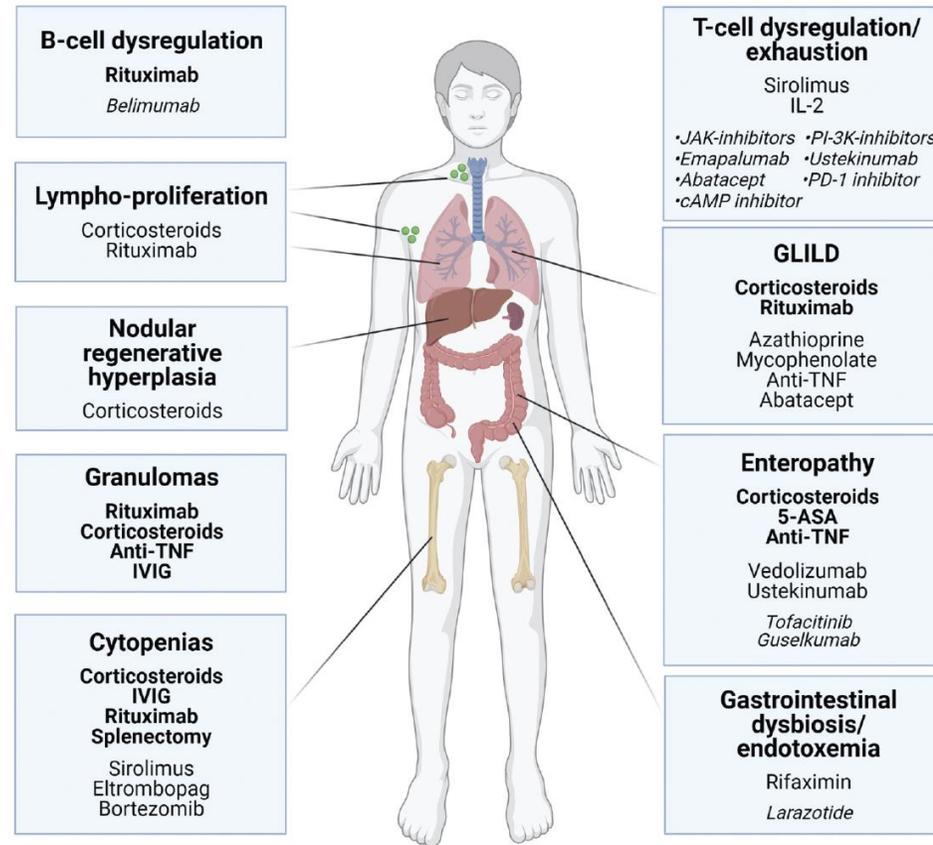
and many others...

Targeted therapies: repurposing drugs

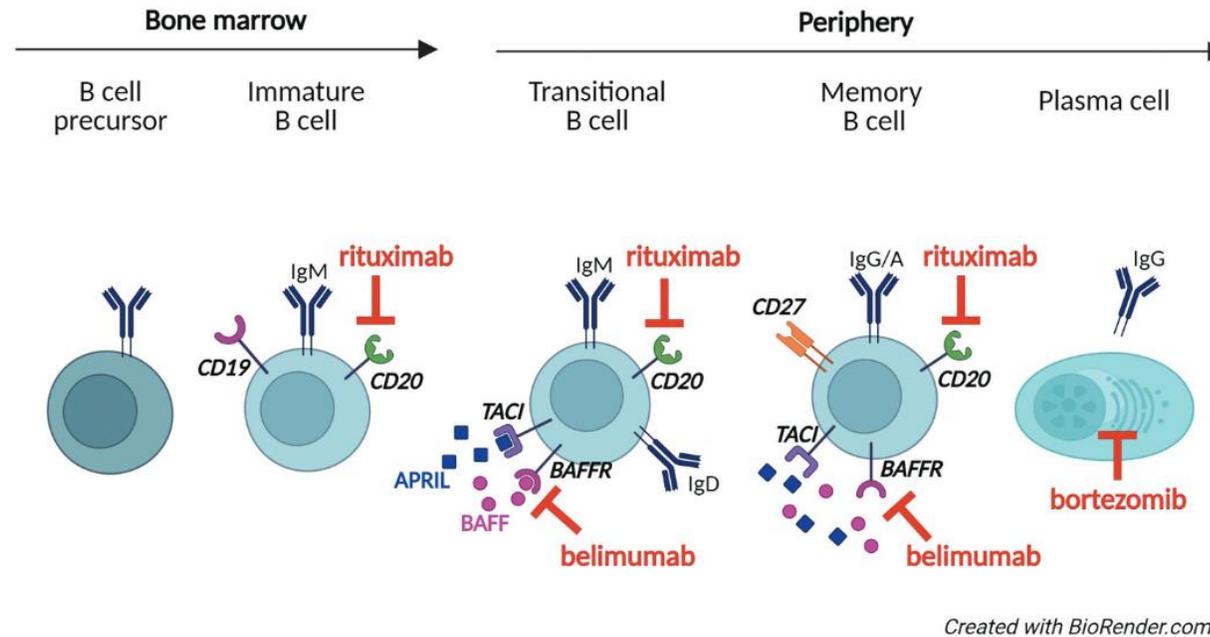


In 2022:
> 800 diff mAb

Targeted therapies: CVID as a good example



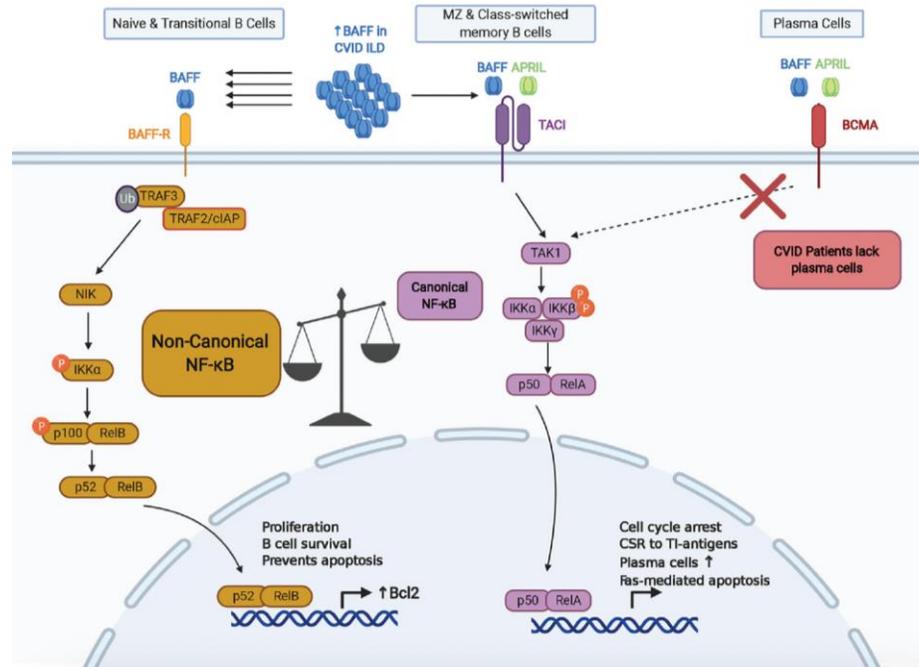
Targeted therapies: CVID as a good example



Anti-BAFF antibody in the form of belimumab could be an alternative or even a supplement to rituximab

Targeted therapies: CVID as a good example

Belimumab?



Developmental and functional abnormalities of B cell compartments observed in CVID ILD suggest that imbalance of B cell signaling networks may promote lung disease.

Included within these potential mechanisms of disease is B cell activating factor (BAFF), a cytokine that potently influences B cell activation and survival

Targeted therapies: the increasing role of JAK-STAT inhibitors

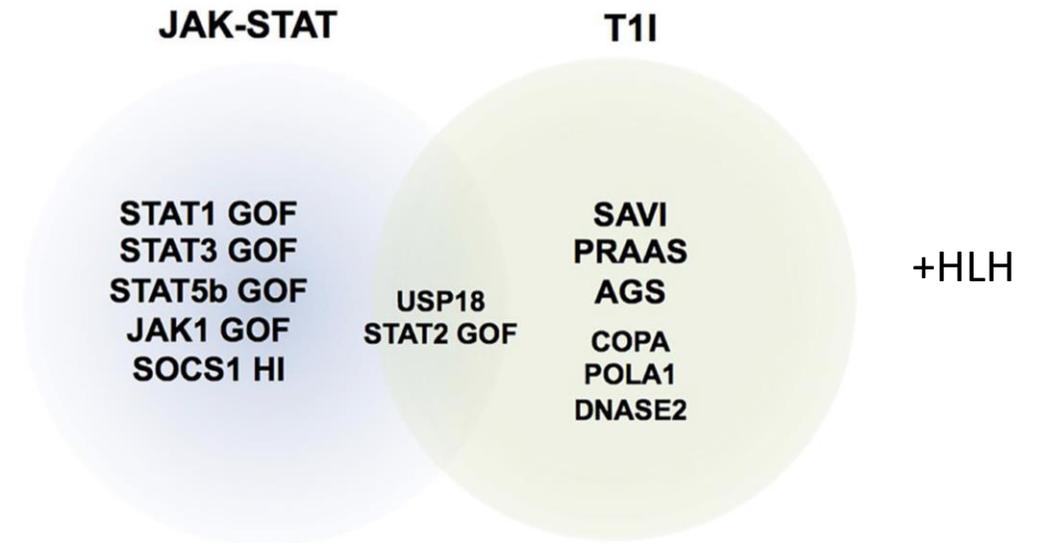
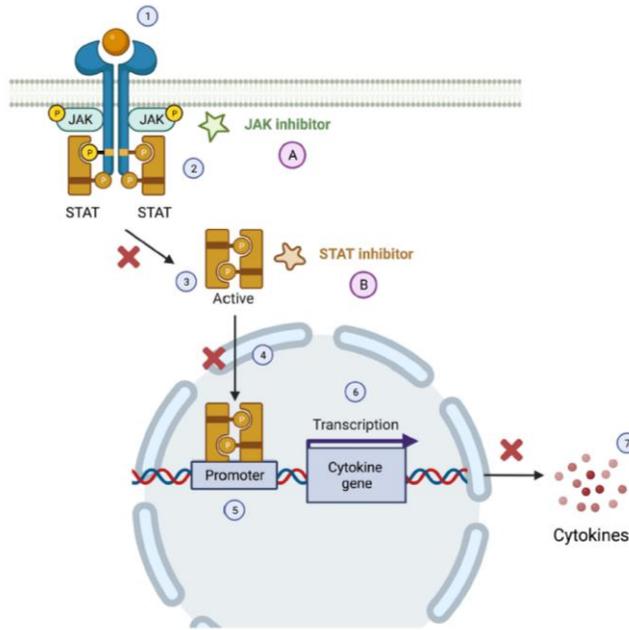


FIGURE 1 | Representative schematic of inborn errors of immunity (IE) where JAK inhibitors have been used. T1I, type I interferonopathies; JAK-STAT, IELs related to mutations in components of the JAK-STAT pathway. HI, haploinsufficiency.

TABLE 2 | Approved marketed JAKinibs.

Name	Specificity	Approved indications	Elimination
Tofacitinib	JAK1/JAK3/(JAK2)	RA, PsA, UC, pA JIA	metab. by Cyto.
Baricitinib	JAK1, JAK2	RA	urine excretion
Ruxolitinib	JAK1, JAK2	MPN, acute GVHD	metab. by Cyto.
Peficitinib	pan-JAK	RA (Japan)	metab. indt of cyto
Fedratinib	JAK2, FIt3	MPN	metab. by Cyto.
Upadacitinib	JAK1	RA, PsA	metab. by Cyto.
Filgotinib	JAK1	RA (Europe, Japan)	urine excretion

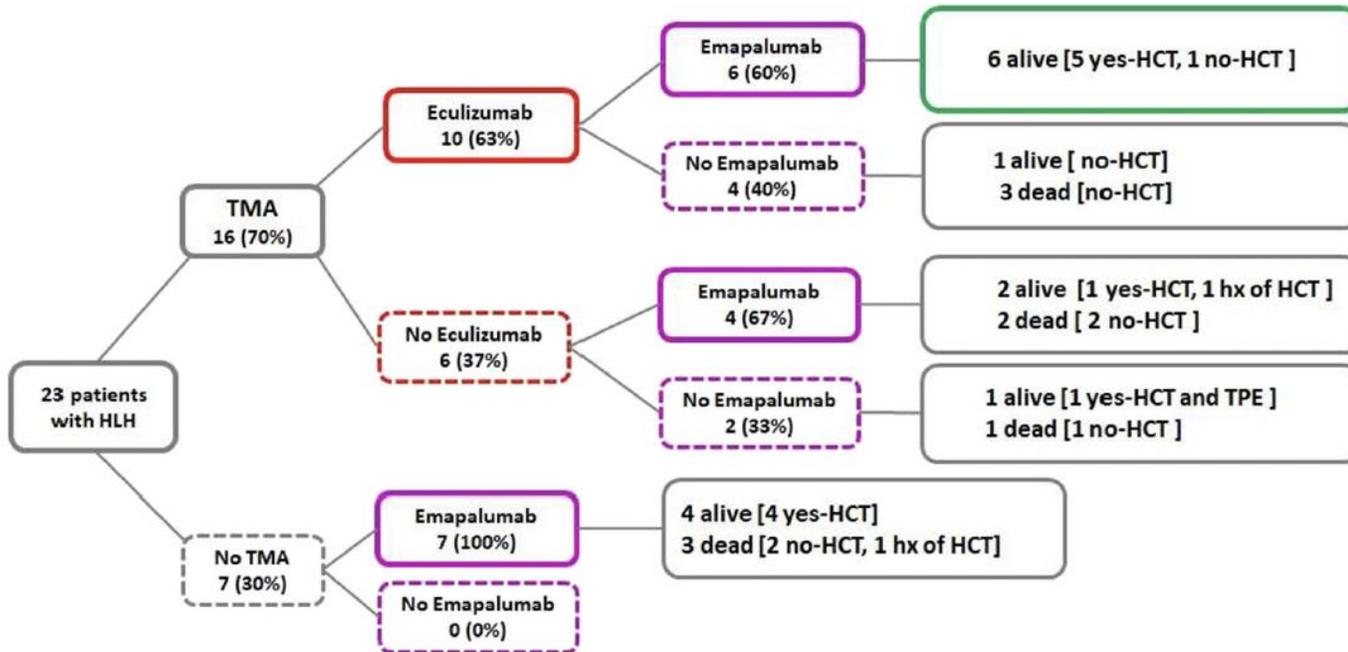
Targeted therapies: the increasing role of JAK-STAT inhibitors

Some open questions:

- What are the risk of infections in the short and long term?
- What is the long-term safety of JAKinibs in children, particularly with regard to growth and bone metabolism?
- Is it a long-term treatment or a bridge to transplant?
- What dosage and what administration scheme should be proposed according to the pathology and the age of the patient?
- Should the benefit of the treatment be monitored clinically and/or biologically, and what are the best readouts?

Targeted therapies: eculizumab in HLH with MAT

Thinking Beyond HLH: Clinical Features of Patients with Concurrent Presentation of Hemophagocytic Lymphohistiocytosis and Thrombotic Microangiopathy



Dual targeted therapy with different targets may also be an option

Is there a role for microbioma modification?

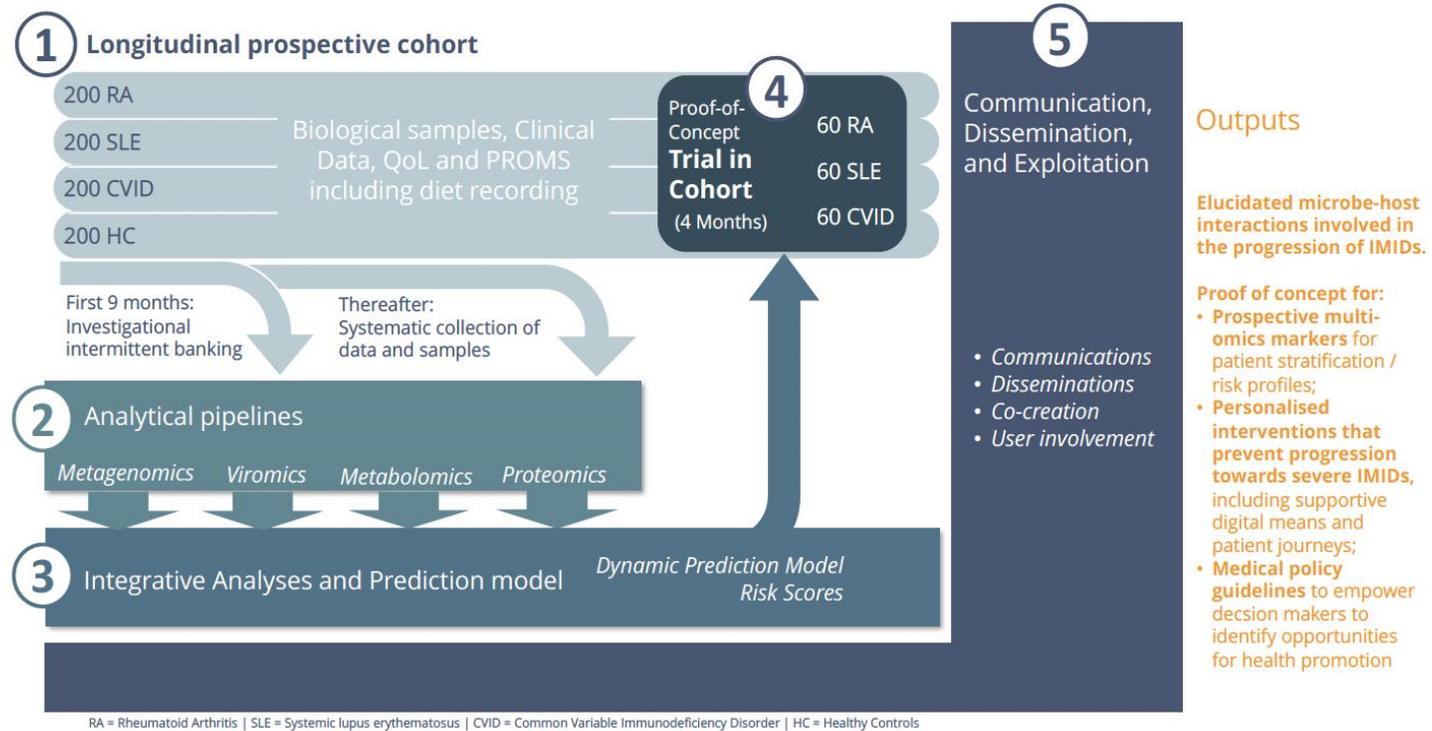
GI dysbiosis as a cause of immune dysregulation and inflammation

- ❑ CVID patients with inflammatory complications have a gastrointestinal dysbiosis compared to CVID patients with infection only and healthy controls.
- ❑ This dysbiosis was related not only to endotoxemia but also to high levels of the inflammatory T-cell marker soluble CD25.
- ❑ In a randomized controlled study, **rifaximin** led to a change in the composition of the gut microbiota in treated patients, but not in the levels of the inflammatory markers

Is there a role for microbioma modification?

**IMIDIATE: PREVENT IMMUNE MEDIATED INFLAMMATORY DISORDERS:
INTERVENTIONAL ALIMENTARY TRIAL IN EUROPE**

#@APP-FORM-HERIAIA1ST@# #@REL-EVA-RE@#



To sum up

- ❑ Gene therapy has potential benefits over SCT, but it is still a limited option for some severe entities
- ❑ Modified haplo-SCT has open the door to transplant all susceptible patients with good results
- ❑ Targeted therapies are real therapeutic options but there are still some unanswered questions with their use
- ❑ Functional studies are useful to detect candidate patients despite no molecular diagnosis and to monitor response to targeted therapies

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Perurena,
Manuel Hernández

Research lab: Marina Garcia, Alba Parra

Inmunogenetics: Roger Colobran, Laura Batlle

VH-IEI Committee



Q & A

COLLABORATION



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Transición de la atención pediátrica a la adulta

Transition care from paediatric to adult clinics

Debate interactivo con un médico de pediátrica y otro de adultos

Interactive panel discussion with a children and an adult physician

Dr Miguel Galicchio, Argentina, y Dra Leila Ferreyra Mufarregue, Argentina.

Dr Miguel Galicchio. Argentina, and Dr Leila Ferreyra Mufarregue, Argentina.

1- QUÉ ES LA TRANSICIÓN?

WHAT IS TRANSITION?

2- HAY UNA EDAD DETERMINADA PARA LA TRANSICIÓN Y CUÁLES SERÍAN LOS PASOS A SEGUIR PARA UNA TRANSICIÓN ADECUADA?

THERE IS A CERTAIN AGE FOR THE TRANSITION AND WHAT WOULD BE THE STEPS TO FOLLOW FOR A PROPER TRANSITION?

**3- QUIÉNES ESTÁN INVOLUCRADOS EN LA TRANSICIÓN?
LAS DIFERENTES ESPECIALIDADES MÉDICAS TAMBIÉN
FORMAN PARTE DE LA TRANSICIÓN?**

**WHO ARE INVOLVED IN THE TRANSITION?
THE DIFFERENT MEDICAL SPECIALTIES ARE ALSO PART OF
THE TRANSITION?**

4- CUÁL ES EN DEFINITIVA EL OBJETIVO DE HACER UNA TRANSICIÓN?

WHAT IS ULTIMATELY THE OBJECTIVE OF THE TRANSITION?

5- QUÉ DIFERENCIAS EXISTEN ENTRE LA ATENCIÓN PEDIÁTRICA Y LA DE ADULTOS?

WHAT DIFFERENCES EXIT BETWEEN PEDIATRIC AND ADULT CARE?

6- CUÁNDO ES EL MOMENTO INDICADO PARA REALIZAR LA TRANSICIÓN Y QUIÉN DECIDE EL LUGAR DE ESA TRANSICIÓN?

WHEN IS THE RIGHT TIME TO MAKE THE TRANSITION AND WHO DECIDES THE PLACE OF THAT TRANSITION?

7- CÓMO SE SABE CUANDO EL PACIENTE ESTÁ PREPARADO PARA LA TRANSICIÓN?

HOW TO KNOW WHEN THE PATIENT IS READY FOR THE TRANSITION?

8- CUÁLES SERÍAN LAS RESPONSABILIDADES Y OBLIGACIONES DEL PACIENTE EN LA TRANSICIÓN?

WHAT WOULD BE THE RESPONSABILITIES AND OBLIGATIONS OF THE PATIENT IN THE TRANSITION?

9- CUÁN IMPORTANTE ES LA AUTONOMÍA Y ADHERENCIA DEL PACIENTE EN LA TRANSICIÓN?

HOW IMPORTANT IS THE PATIENT'S AUTONOMY AND ADHERENCE IN THE TRANSITION?

10- EXISTEN OBSTÁCULOS EN LA TRANSICIÓN?

THERE ARE OBSTACLES IN THE TRANSITION?

11- CÓMO ES LA TRANSICIÓN EN LA ARGENTINA?

HOW IS THE TRANSITION IN ARGENTINA?

12- CÓMO PUEDEN AYUDAR LAS ORGANIZACIONES DE PACIENTES EN EL PROCESO DE TRANSICIÓN O EN SU IMPLEMENTACIÓN?

HOW PATIENT ORGANIZATIONS HELP IN THE TRANSITION PROCESS OR ITS IMPLEMENTATION?

13- PORQUÉ ES TAN COMPLEJO IMPLEMENTAR LA TRANSICIÓN EN TODOS LOS PAÍSES?

CUÁL SERÍA LA CAUSA?

**WHY IS IT SO COMPLEX TO IMPLEMENT THE TRANSITION IN ALL COUNTRIES?
WHAT WOULD BE THE CAUSE?**

**THANKS
MUCHAS GRACIAS!**

COLLABORATION



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Q & A

COLLABORATION



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Compartiendo el éxito: Acciones impactantes de las NMOs en diagnóstico, tratamiento y cuidados

Sharing Success: NMOs' impactful actions in diagnosis, treatment, and care

Miriam Ferreira, Responsable de programas de las NMOs de IPOPI
Miriam Ferreira, IPOPI NMO Programmes Officer



ARGENTINA

Argentina - Asociación Civil Ayuda al Paciente con Inmunodeficiencia Primaria. 2022

Challenge:

- Implement a simple and effective tool for patients and their families where they can find safe and accurate information about their doubts and concerns so that they do not search the Internet for erroneous information.

Action summary:

- Available in APP STORE and PLAYSTORE
- Application with 10 categories. Disability certificate, Diagnosis, adverse effects, Rare diseases, Primary immunodeficiencies, Myths, Family planning, Plasma, Treatments and Access.
- 1-minute explanatory videos made by professionals specializing in each of the disciplines.
- It also gives us the possibility of surveying topics of interest to patients through the search engine and thus generating new content.

Results:

- From December 2022 to September 2023, 1.853 users registered.
- 53 users registered to have the possibility to make suggestions of new topic.



Ranking preguntas		
Puesto	Pregunta	Cantidad vistas
1	¿Qué hacer si durante una infusión de gamaglobulina tengo fiebre?	101
2	¿Todas las IDP pueden acceder al CUD?	52
3	Enfermedades poco frecuentes	40
4	Tienen cura las IDP	29
5	¿Cuáles son las indicaciones de inmugamaglobulinas subcutánea y endovenosa?	27
6	¿Por qué algunas IDP reciben gamaglobulina y otras no?	20
7	¿Por qué las OS nos piden el CUD? Parte 1	16
8	¿Cuáles son los efectos adversos de la gamaglobulina?	15
9	¿Cómo disminuir o eliminar el dolor de cabeza por 24hs que me produce la gamaglobulina endovenosa?	14

Argentina - Asociación Civil Ayuda al Paciente con Inmunodeficiencia Primaria. 2022

Desafío:

- Implementar una herramienta simple y efectiva para los pacientes y sus familias en donde puedan encontrar información segura y certera sobre sus dudas e inquietudes para que no busquen en internet con información errónea.

Resumen de la acción:

- Disponible en APP STORE y PLAYSTORE
- Aplicación con 10 categorías. CUD, Diagnóstico, Efectos adversos, Enfermedades poco frecuentes, Inmunodeficiencias Primarias, Mito, Planificación familiar, Plasma y Tratamientos, Acceso.
- Videos explicativos de 1 minuto realizados por profesionales especialistas en cada una de las disciplinas.
- Nos brinda también la posibilidad de relevar los temas de interés de los pacientes a través del buscador y así generar nuevos contenidos.

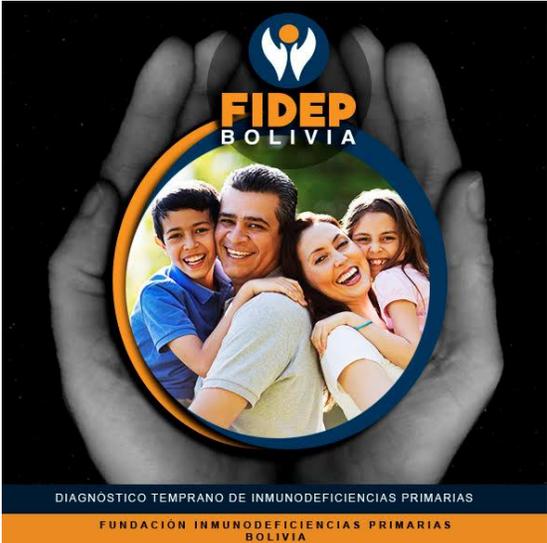
Resultados:

- Desde Diciembre 2022 al Septiembre 2023 han ingresado 1.853 usuarios mensuales
- Se registraron 53 usuarios para poder tener la posibilidad de enviar sugerencias de nuevos temas.

Ranking preguntas

Puesto	Pregunta	Cantidad vistas
1	¿Qué hacer si durante una infusión de gamaglobulina tengo fiebre?	101
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9	¿Cómo disminuir o eliminar el dolor de cabeza por 24hs que me produce la gamaglobulina endovenosa?	14





BOLIVIA

FIDEP BOLIVIA

Challenge

- 1. From 2019. Achieve free access to Immunoglobulins for all PID patients. Immunoglobulins should be included in the National List of Essential Medicines.**
- 2. In 2023: Remove from the market an Immunoglobulin from India that gave very strong side effects to all patients (fevers, convulsions, etc.).**

Action summary

- 1. Meetings between our team of immunological and legal advisors from FIDEP with the Ministry of Health and MPs, demonstrating the need and importance.**
- 2. Medical reports of side effects from patients to the drug agency. A lot of pressure from relatives.**

Results

- 1. 2022: We successfully achieved access to immunoglobulins for all our patients.**
- 2. The drug agency took the India Immunoglobulin off the market.**

FIDEP BOLIVIA

Desafío

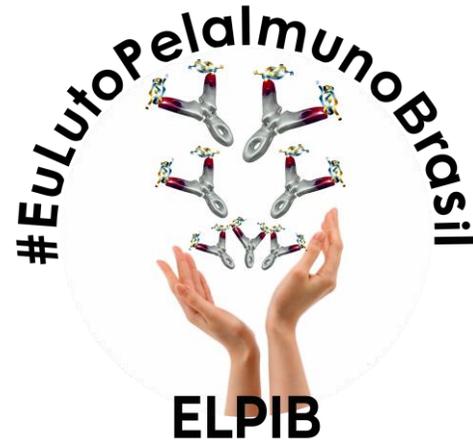
- 1. Desde el año 2019. Conseguir acceso gratuito a las Inmunoglobulinas para todos los pacientes con IDP. Que las Inmunoglobulinas estén dentro del Listado Nacional de Medicamentos como esenciales.**
- 2. Año 2023. Sacar del mercado una Inmunoglobulina de la India que dio efectos secundarios muy fuertes a todos los pacientes. (fiebres, convulsiones, etc)**

Resumen de la acción

- 1. Reuniones de nuestro equipo de asesoras inmunólogas y legales de FIDEP con el Ministerio de Salud y Diputados, demostrando la necesidad e importancia.**
- 2. Reportes médicos de efectos secundarios de los pacientes a la agencia del medicamento. Mucha presión de los familiares.**

Resultados

- 1. Año 2022. Conseguimos con mucho éxito el acceso a las Inmunoglobulinas para todos nuestros pacientes.**
- 2. La agencia del medicamento sacó la Inmunoglobulina de la India del mercado.**



BRAZIL

Challenge:

Combat underdiagnosis of primary immune deficiency by raising awareness among medical residents warning signs across various specialties.

Summary of the action:

Medical residents from Federal University of São Carlos-UFSCAR and Teaching and Research Institute of Santa Casa de Misericórdia de São Carlos-IEP were invited to a presentation by UFSCAR immunology professor Dra Flávia Pileggi Gonçalves with the titled "When to suspect innate immunity errors in different medical specialties"

Outcomes:

Participation of 92 doctors at the presentation, followed by the celebration dinner.

Desafío:

Combatir el infradiagnóstico de las Inmunodeficiencias primarias mediante la sensibilización de los estudiantes de medicina mediante los signos de alarma de diferentes especialidades.

Resumen de la acción:

Los estudiantes de medicina de la Universidad Federal de São Carlos - UFSCAR y el Instituto de Enseñanza e Investigación de la Santa Casa de Misericórdia de São Carlos- IEP fueron invitados a una presentación por parte del professor de inmunología de UFSCAR Dra Flávia Pileggi Gonçalves com el título “Cuando sospechar de un error de la inmunidad innata en diferentes especialidades médicas”

Resultados:

Participación de 92 doctores en las presentaciones seguidas de una cena de celebración..

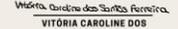


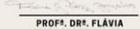
CSL Behring
Biotherapies for Life™



CERTIFICADO

Certificamos que *NOME COMPLETO* participou da Palestra:
Quando suspeitar de erros inatos da imunidade nas
diversas especialidades médicas em 26 de abril de 2023 na
cidade de São Carlos - SP como ouvinte.


VITÓRIA CAROLINE DOS SANTOS FERREIRA
 Presidente da Liga de Imunologia e Alergia da UFSCar


PROF. DR. FLÁVIA PILEGGI GONÇALVES
 Orientadora da Liga de Imunologia e Alergia da UFSCar


DR. JUCAIRA STELLA MARTINS GIUSTI
 Vice-presidente da Associação Eu Luto Pela Imunidade Brasileira



COLLABORATION



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CHILE

Ley Ricarte Soto:

¿CÓMO SE ACCEDE?

- El médico tratante se debe inscribir en el Sistema informático de la ley Ricarte Soto disponible en la Web del Fonasa, para efectuar la postulación del paciente.
- Será el médico tratante: Inmunólogo, Internista o Pediatra, quien deberá realizar la postulación del beneficiario a este Sistema* en la plataforma web del Fonasa.
- El médico tratante será el responsable de aportar de manera íntegra y oportuna toda la información requerida, asegurando la correcta postulación del beneficiario.
- Comité de Expertos Clínicos del Prestador Aprobado debe evaluar inclusión del beneficiario.

BENEFICIO LEY RICARTE SOTO

IDP

Atención que **GARANTIZA**

- No garantiza, examen confirmación diagnóstica
- Sí incluye medicamento

¿DÓNDE SE ENTREGA?

Un médico tratante y/o Fonasa le informará sobre el prestador* que entregará el beneficio.

*Estos prestadores de salud, públicos o privados, estarán previamente autorizados por el Ministerio de Salud para este Sistema.



Ley Ricarte Soto:

TIEMPO DE ESPERA

- **Inicio Tratamiento (para pacientes con inmunodeficiencia primaria que cumplan criterios de inclusión):**
- **Inicio Inmunoglobulina G endovenosa: 14 días desde la validación de la indicación por Comité de Expertos Clínicos del prestador aprobado.**
- **Inicio Inmunoglobulina G subcutánea 14 días desde la validación de la indicación por Comité de Expertos Clínicos del prestador aprobado, que presente al menos anafilaxia documentada debida a Inmunoglobulina G endovenosa, síntomas documentados relacionados a la infusión de Inmunoglobulina G endovenosas no manejables y/o imposibilidad certificada de obtener accesos venosos.**
- **Continuidad de atención y control: Según lo establecido en protocolo.**
- **Seguimiento: Según lo establecido en protocolo**

BENEFICIO LEY RICARTE SOTO

IDP

MONTO A PAGAR

- **Medicamento sin costo para el beneficiario (*).**

¿QUÉ PRESTACIONES CUBRE??

- **Inmunoglobulina G endovenosa o subcutánea**



Foundation achievements :



Networking:

- member of IPOPI (International patient organisation for primary Immunodeficiencies) where 2022 attended the international conference on primary immunodeficiencies in Gothenburg, Sweden.
- April 2023 , Lunch with members of the foundation, where new ideas are articulated and relevant information is passed on.
- INICIATIVA ALAS; Participation in 3rd Regional Meeting in CDMX.



Logros de la fundación:

Creación de redes:

- miembro de IPOPI (International patient organisation for primary Inmunodeficiencias) donde el año 2022 se asistió a la conferencia internacional de inmunodeficiencias primarias en Gotemburgo, Suecia.
- Abril 2023 , Almuerzo con miembros de la fundación , donde se articulan nuevas ideas y se transmite información relevante.
- INICIATIVA ALAS; Participación en 3° Reunión Regional en CDMX.





ECUADOR

Ecuador Fundación PIDE

Challenge:

Treatment (Intravenous Immunoglobulin)

Action Summary:

Pharmacovigilance entry of new brand into Ecuador without EMA and FDA approval, without approved clinical studies.

Results:

A judicial process is carried out (Autonomous Precautionary Measures), where the sentence is won in favour, in which the Sanitary Registration of the medicine is withdrawn, and its distribution and commercialisation in the country is prohibited due to non-compliance with quality, safety and efficacy guarantees.

Ecuador Fundación PIDE

Desafío:

Tratamiento (Inmunoglobulina Intravenosa)

Resumen de la acción:

Farmacovigilancia ingreso de nueva marca a Ecuador sin aprobación EMA y FDA, sin estudios clínicos aprobados.

Resultados:

Se realiza un proceso judicial (Medidas Cautelares Autónomas), donde se gana a favor la sentencia, en la que se retira el Registro Sanitario del medicamento, se prohíbe su distribución y comercialización en el país por incumplimiento de garantías de calidad seguridad y eficacia.



ECUADOR

FUNDACIÓN PIDE



ACUERDO MINISTERIAL
No. 00005-2021



GRACIAS

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EL SALVADOR

El Salvador – Escudo de Amor para las IDP

Challenge:

Impossibility to obtain a genetic diagnosis of an IDP at El Salvador

Summary of the action:

Members of El Salvador Organizacion (doctors) obtained an agreement with the laboratory INVITAE for the shipment and Processing of samples, they lead the family patients through the diagnosis process.

Outcomes:

Accurate genetic diagnosis.

Specific treatment for the type of condition.

Clarity for the family regarding the origin of the condition and treatment.

Advice and follow up to the family.

El Salvador – Escudo de Amor para las IDP

Desafío:

Imposibilidad de obtener diagnóstico Genético en El Salvador

Resumen de la acción:

Miembros de la Fundación (médicos) obtuvieron acuerdo con el laboratorio INVITAE para envío y procesamiento de muestras.

Resultados:

Diagnóstico genético oportuno

Tratamiento específico para el tipo de padecimiento

Claridad para la familia en cuanto al origen del tratamiento y padecimiento

Asesoría y seguimiento a la familia

MEXICO

PARAGUAY

Paraguay (ASPANA)

Challenge:

Through educational tools and random controls with medical specialists responsible for the USF (units for family Health). The challenge is to detect patients with primary immunodeficiencies.

Summary of the action:

- 1. A call to managers of USF and, through them, to paediatricians and clinical doctors.*
- 2. We select a date for the for the educational talks (primarily with professionals and then with patients).*
- 3. In a future visit, medical checks will be done and detection of potential patients with PIDs according to signs and symptoms and then they make the necessary studies.*

Outcome:

1. Between 2021-2023, 56 patients with PIDs (children and adults) were identified.
2. They continue with their treatment, with the help of ASPANA (to date, it is mainly sustained on the basis of activities developed by parents of patients: clothes fairs, food fairs, etc).

Paraguay (ASPANA)

Desafío:

Atreves de charlas educativas y controles aleatorios con médicos responsables de las USF (unidad de salud familiar). El desafío es lograr detectar pacientes con inmunodeficiencias.

Resumen de la acción:

- 1. Se hace un llamado a los directivos de las USF y atreves de ellos a médicos pediatras y médicos clínicos.*
- 2. Marcamos una fecha para las charlas educativas (primeramente con los profesionales y luego con los pacientes).*
- 3. En una próxima visita se realizan los controles médicos y se hacen detecciones de probables pacientes con inmunodeficiencias según signos y síntomas y luego se realizan los estudios necesarios.*

Resultado:

1. Se lograron detectar entre el 2021-2023, 56 pacientes con inmunodeficiencias entre niños y adultos.
2. Siguen sus tratamientos, con la ayuda de la ASPANA (que hasta el día de hoy se sustenta con las actividades que realizan los padres de la Asociación (feria de ropas, feria de comidas, entre otros).



PUERTO RICO

Puerto Rico - APIP

Challenge:

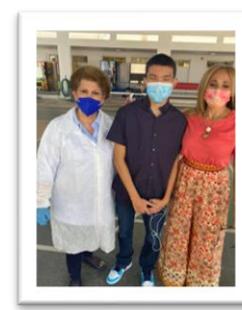
Get patients with PIDs in Puerto Rico involved with their diagnosis and help achieve progress with the different sectors: government, Health insurers, Health professionals, community environment that results in short and long-term benefits for everyone

Summary of the action:

- *Early access to COVID vaccine – August 2021*

Outcomes:

- *In the middle of the pandemic, APIP succeeded in getting the PID patients and their families vaccinated.*
- *In August 2021, more than 175 persons of our PID patient community benefited from this action.*
- *We were the only patient association that organised an action like this one in a context of Shortages of COVID vaccines.*
- *We prepared a press release, the media in the country helped in spreading the word about the action and provided media coverage to the evento.*
- *We worked in the security and hygienic protocols that the evento and the circumstances required.*



Puerto Rico - APiP

Desafío:

Lograr que los pacientes de IDP en Puerto Rico se involucren con su diagnóstico y ayuden a lograr avances con los diferentes sectores: Gobierno, aseguradoras médicas, profesionales de la salud, entorno comunitario en general que redunden en beneficio a corto y a largo plazo para todos.

Resumen de la acción:

- *Acceso precoz a la vacuna del COVID - agosto 2021*

Resultados:

- *En plena pandemia APiP gestionó la vacunación de nuestra comunidad y sus familiares*
- *El agosto del 2021 más de 175 personas de nuestra comunidad de pacientes con IDP se beneficiaron de esta acción*
- *Fuimos la única organización de pacientes que organizó una acción como esta dentro de la escasez de las vacunas del COVID*
- *Se trabajó comunicado de prensa, los medios de comunicación del país nos ayudaron a difundir el aviso y cubrieron el evento*
- *Trabajamos en los protocolos de seguridad e higiene que el evento y las circunstancias ameritaban*





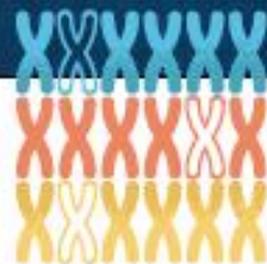
URUGUAY

DESAFÍOS

- *Centralizar a los pacientes
- *Concientizar a la población sobre los signos de alarma
- *Con ese fin, los médicos a cargo del grupo de inmunodeficiencias generaron instancias con la finalidad de capacitar a los médicos y ponerlos en alerta ante las alarmas por sospecha de estas enfermedades.



TRATAMIENTOS Y LOGROS



*Sé pudo empezar a acceder a los estudios genéticos

*Ahora en el medio local tenemos secuenciadores NGS (técnica next generation sequencing) con el equipo Illumina, en la en la fundación Pérez scremini.

*Contamos con el respaldo de la Jeffrey Modell a través de un convenio que cubre 10 estudios con el panel de inmunodeficiencia al año, acción que facilita enormemente el diagnóstico.



TRABAJO EN EQUIPO

SE FORMÓ UN GRUPO SÓLIDO COMPUESTO POR UN EQUIPO MÉDICO COMPROMETIDO Y TAMBIÉN PACIENTES Y PADRES DE PACIENTES.



RESULTADOS LOGRADOS

CAPACITACIÓN

QUE SE ESTÉN HACIENDO CURSOS DE CAPACITACIÓN PARA PEDIATRAS Y TAMBIÉN PARA ADULTOS ES UN GRAN PASO.

EXTENDIENDO REDES

SE FORMÓ TAMBIÉN UN GRUPO DE ADULTOS QUE TRABAJA EN CONJUNTO SOBRE TODO PARA FACILITAR LA TRANSICIÓN DE PACIENTE PEDIÁTRICO A ADULTO.





EN RESUMEN...

Aún se necesitan muchos recursos especialmente porque en Uruguay no existe el posgrado de inmunología.

Todos quienes integran este equipo tuvieron que salir y estudiar afuera para formarse y capacitarse.

Por eso quisiera destacar especialmente a este grupo de médicos que se dedica de forma incansable para lograr que se aborden estas enfermedades



CONCLUSIÓN CONCLUSION

Roberta Anido de Pena

ALMUERZO LUNCH 1:30 H

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