

XXXI CONGRESO
INTERNACIONAL



XXXI CONGRESO INTERNACIONAL **SCAI 2025**

SOCIEDAD CHILENA DE ALERGIA E INMUNOLOGÍA

MANEJO DE MANIFESTACIONES NO INFECCIOSAS DE INMUNODEFICIENCIA EN EL ADULTO

Dra. Martina Meier
Inmunóloga clínica

Red UC-Christus
Fundación Arturo López Pérez
Centro del Alérgico

AGENDA

Autoinmunidad y desregulación

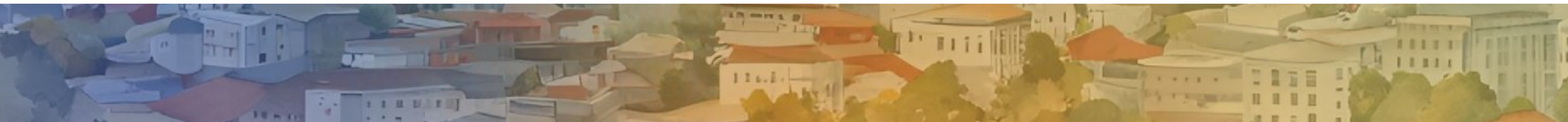
Manifestaciones respiratorias

Manifestaciones digestivas

Linfoproliferación y cáncer

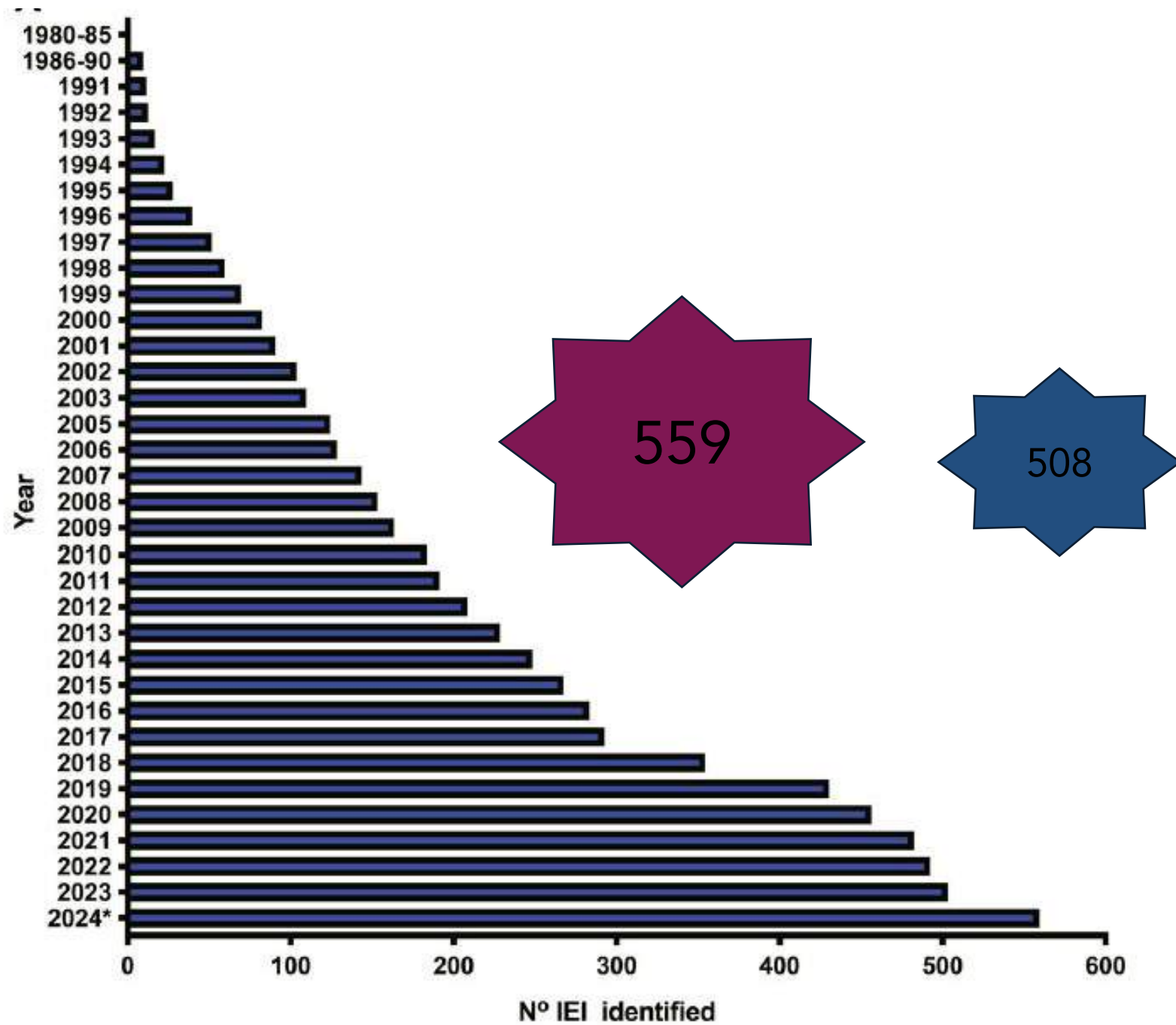
Riesgo cardiovascular

Salud mental



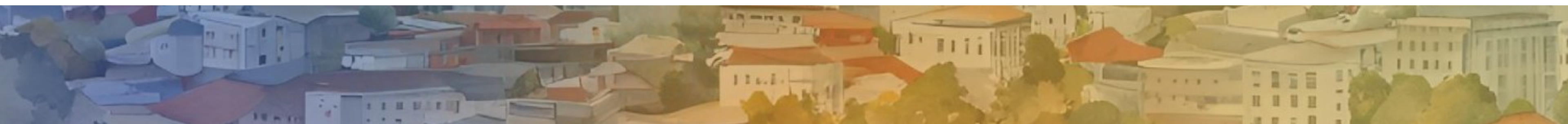
Human inborn errors of immunity: 2024 update on the classification from the International Union of Immunological Societies Expert Committee

1990
(1970)

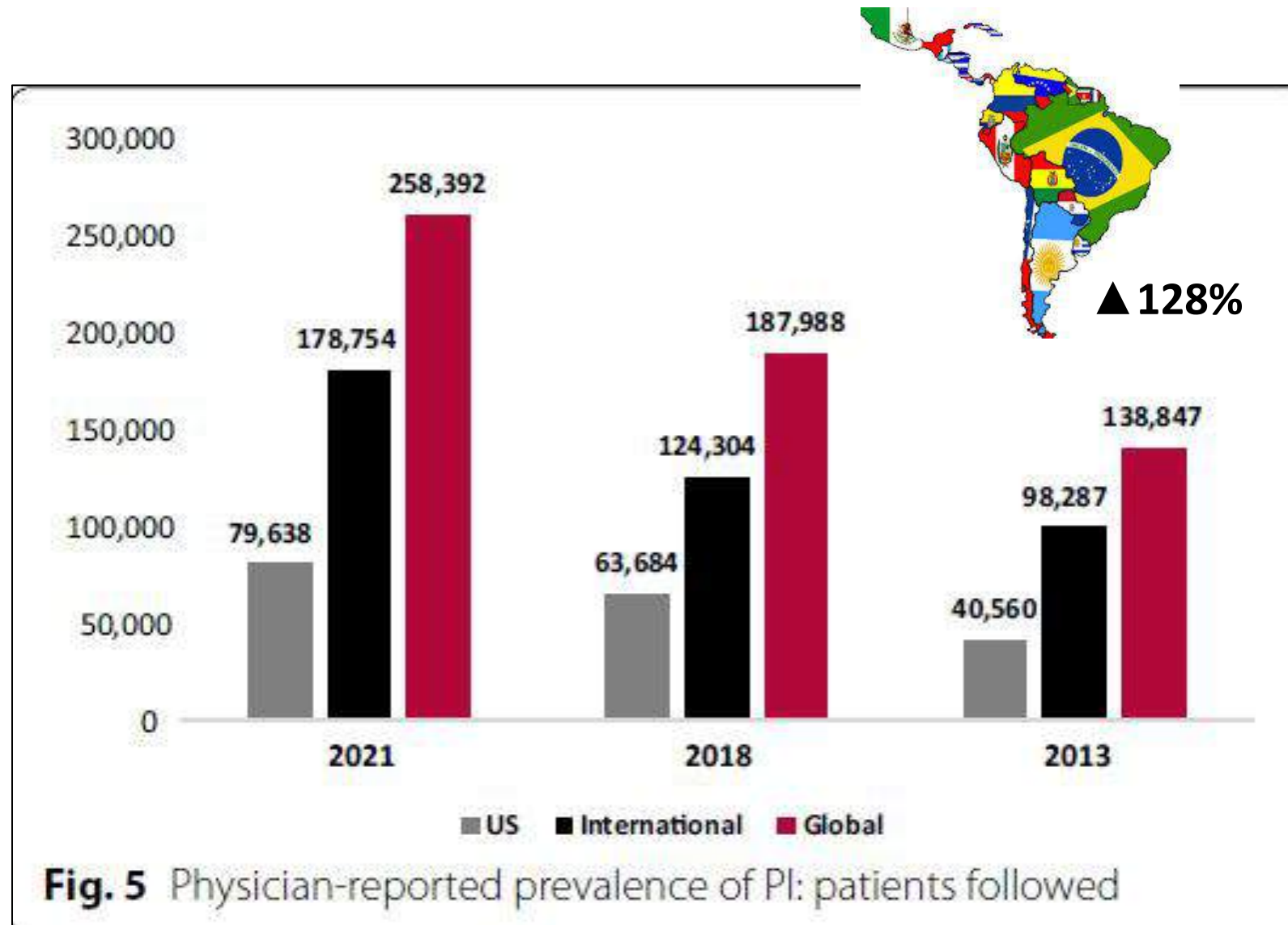


2017

2024



¿ENFERMEDADES RARAS?



6/10.000
(2024)



0.1 - 1%

50% >25 años

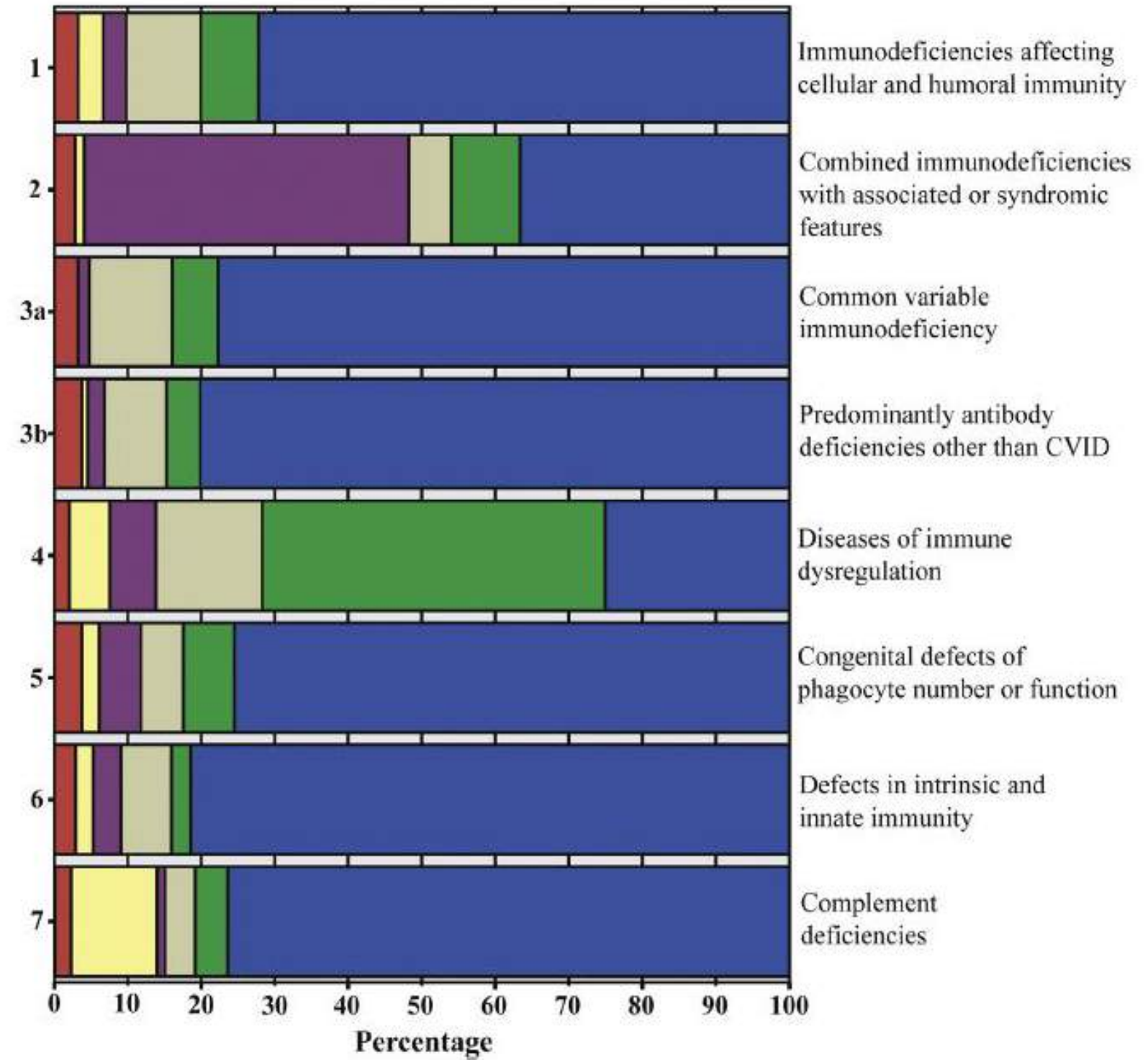
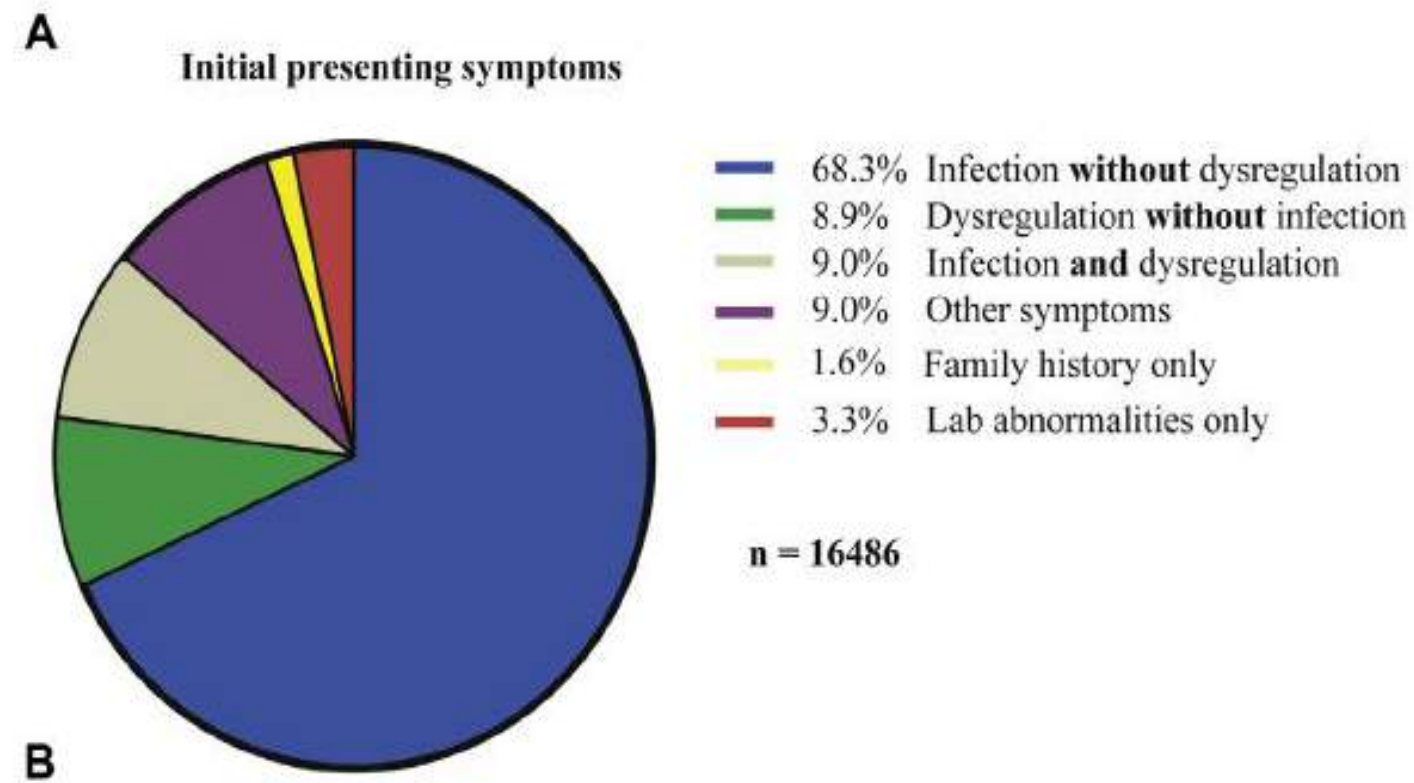
Retraso 16 años

Rider N et al. J Allergy Clin Immunol. 2024 Jun;153(6):1704-1710. doi: 10.1016/j.jaci.2024.01.011 a
Quinn J et al. Allergy Asthma Clin Immunol. 2022 Mar 4;18(1):19. doi: 10.1186/s13223-022-00662-6
Mengyue D et al. BMJ Paediatr Open. 2023 Jul;7(1):e002002. doi: 10.1136/bmjpo-2023-002002

Initial presenting manifestations in 16,486 patients with inborn errors of immunity include infections and noninfectious manifestations

Check for updates

Julian Thalhammer, MD,^{a,b,*} Gerhard Kindle, MD,^{a,*} Alexandra Nieters, PhD,^a Stephan Rusch,^a Mikko R. J. Seppänen, MD,^c Alain Fischer, MD,^{d,e} Bodo Grimbacher, MD,^{d,f,g,h} David Edgar, FRCP, FRCPath,^{i,j} Matthew Buckland, MD,^{k,l} Nizar Mahlaoui, MD, MPH, PhD,^{d,o} and Stephan Ehl, MD,^{a,g} for the European Society for Immunodeficiencies Registry Working Party†
 Freiburg, Germany; Helsinki, Finland; Paris, France; Dublin, Ireland; and London, United Kingdom



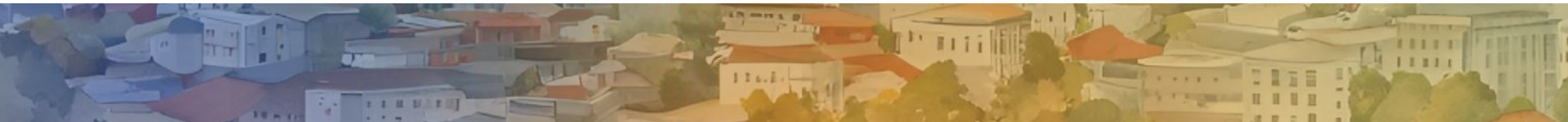
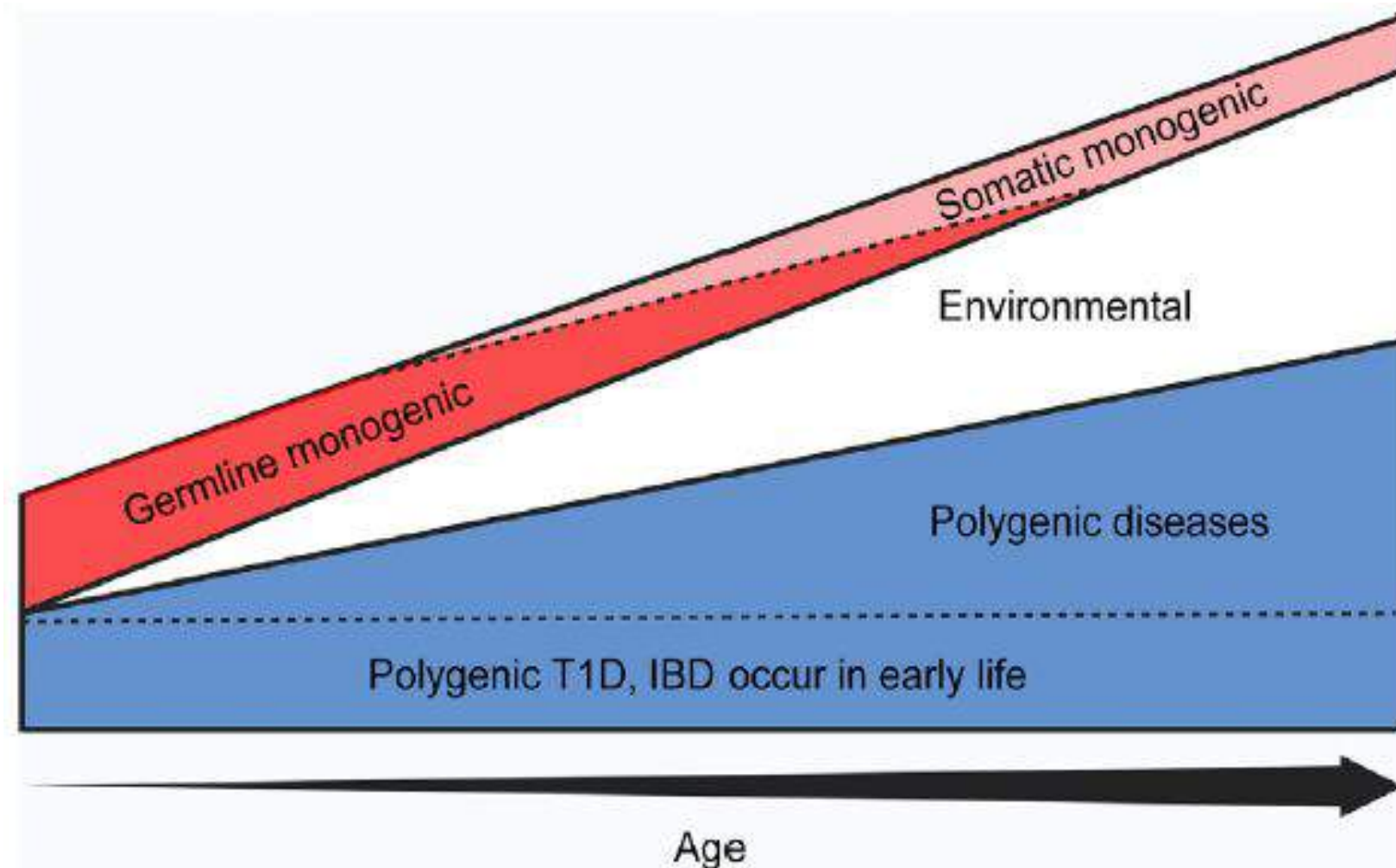


AUTOINMUNIDAD

LA AGUJA MONOGÉNICA EN EL PAJAR POLIGÉNICO

Al poligénica: 5%

Variantes genéticas (HLA; SNP); región no codificante; bajo impacto



AUTOINMUNIDAD EN EII

4

Desregulación (130)



Aparición temprana



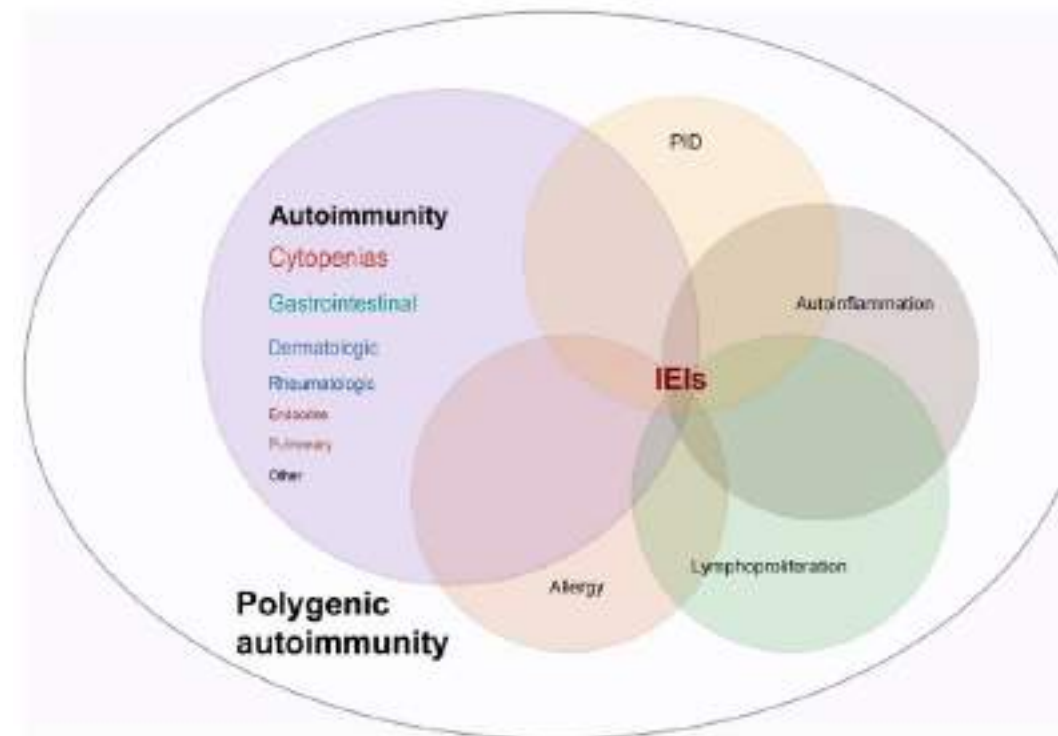
Overlap



Pronóstico

TABLE I. Notable monogenic genes conferring susceptibility to common autoimmune diseases

Autoimmune disease	Notable genes conferring susceptibility
Diabetes mellitus (early-onset)	<i>CTLA4, FOXP3, IL2RA, LRBA, STAT3, STAT1</i>
IBD	<i>ADAM17, FOXP3, IL10, IL10RA, IL10RB, LRBA, MEFV, MVK, SKIV2L,</i> and the groups of CGD genes and atypically presenting SCID genes
Pediatric Evans syndrome (multiple autoimmune cytopenias)	<i>ADAR1, CASP10, CTLA4, FAS, FASL, KRAS, LRBA, NRAS, PIK3CD, RA</i>
SLE	<i>ACP5, CYBB</i> (carrier state), <i>SNRPN, SSB, TMEM173, TREX1,</i>
Arthritis	<i>CD40, CTLA4, FCGR3A, IL23R, IL2RA, IL2RB, IL6ST, COPA</i>
JIA	<i>FAS, IL2RA, IL2RB, IL6, COPA, LACC1</i>



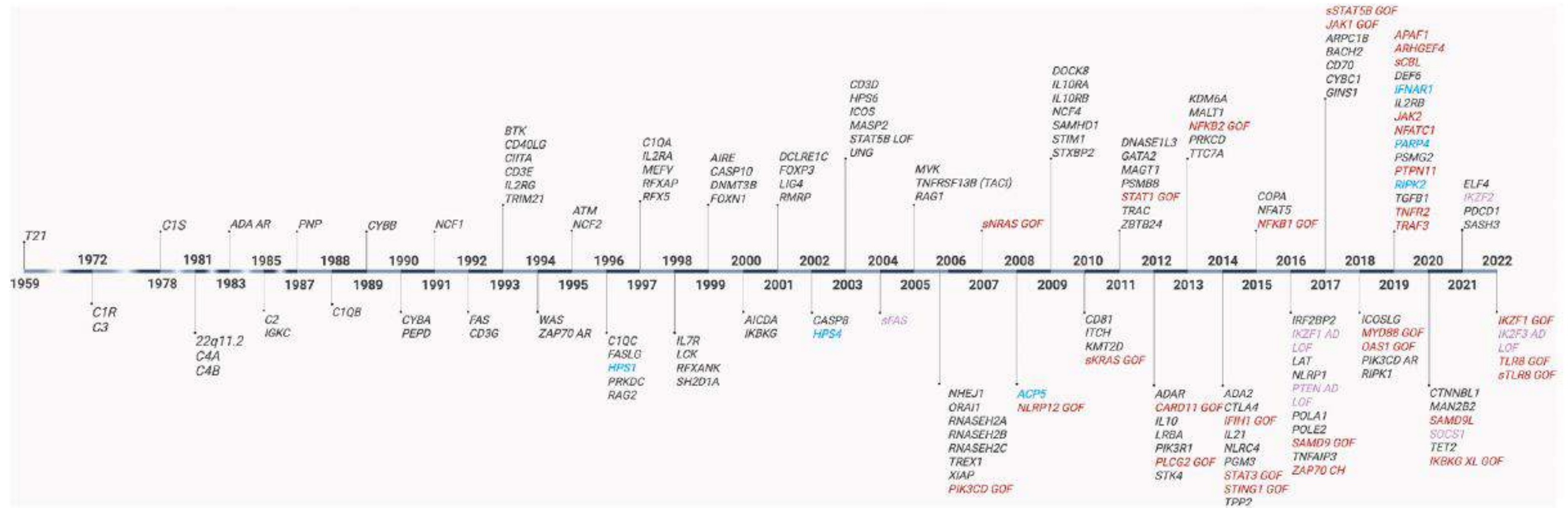


FIGURE 4. Timeline of discovery of PIRDs. Timeline showing the discovery of monogenic PIRDs, which includes genes with a potential autoimmune disease by the date of gene discovery (unless there was a date where a specific genetic entity emerged for a gene that was previously felt to be minimally autoimmune, eg, *STAT3* GOF, *STAT1* GOF, *STAT5b* GOF, *JAK1* GOF, and hypomorphic *CARD11*). CH, Compound heterozygous. Key: Black color: predominantly PID with autoimmunity (including chromosomal anomalies and syndromes); Blue color: recessive autoimmunity; Purple color: germline and somatic heterozygous LOF autoimmunity; Red color: germline and somatic GOF autoimmunity.

AUTOINMUNIDAD EN EII

Tolerancia central
(*AIRE, 22q11d*)

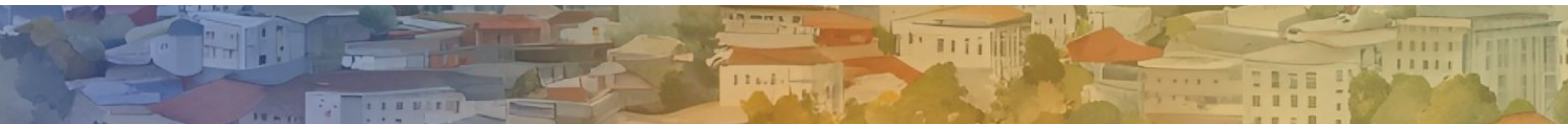
Tolerancia periférica
(*FOXP3, IL2RA*)

Apoptosis
(*ALPS; FAS, CASP*)

Clearance Ag. Propios
(complemento)

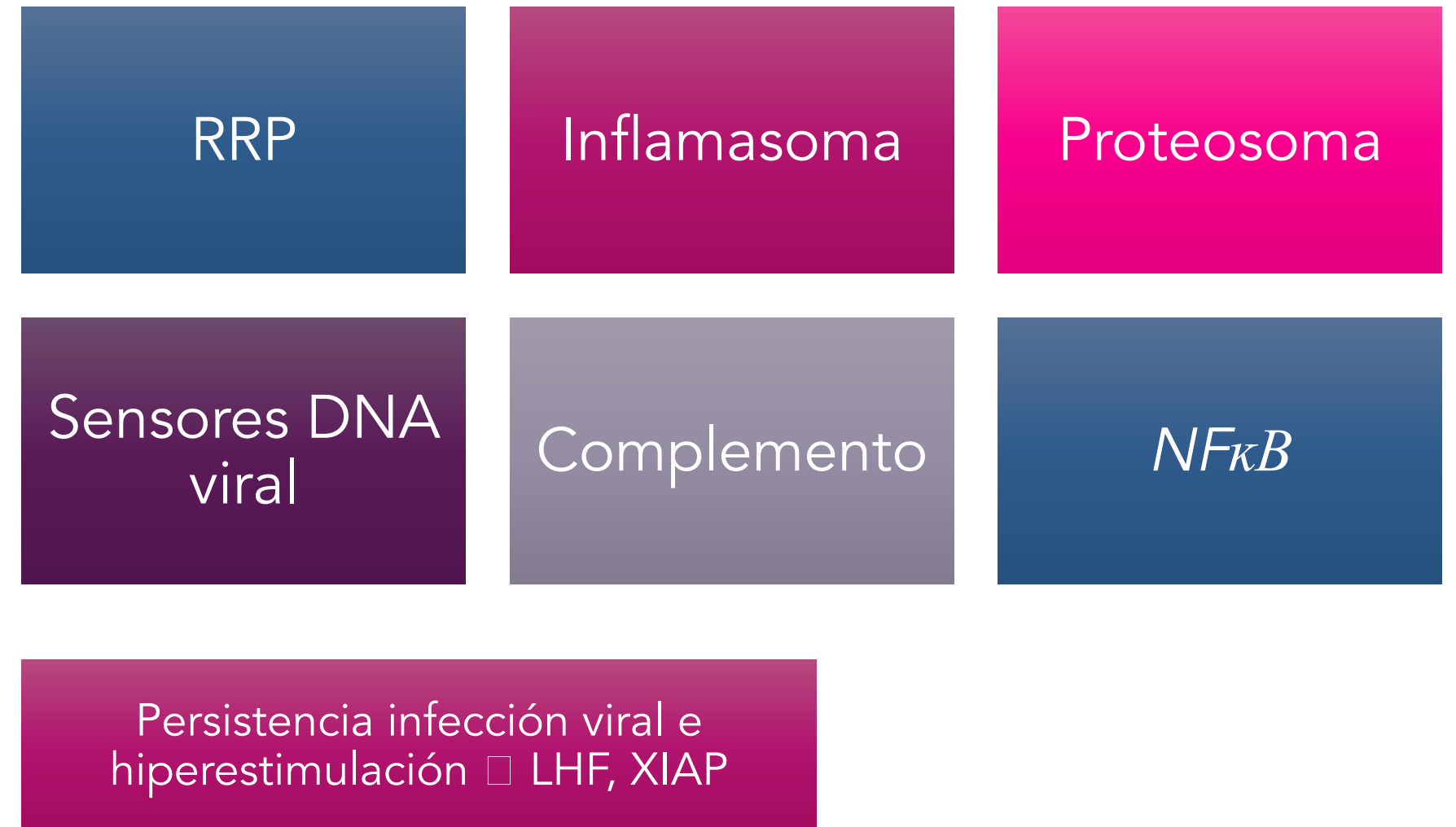
Señalización inhibitoria
(*CTLA4; IL10*)

Señalización activadora
(*STAT*)



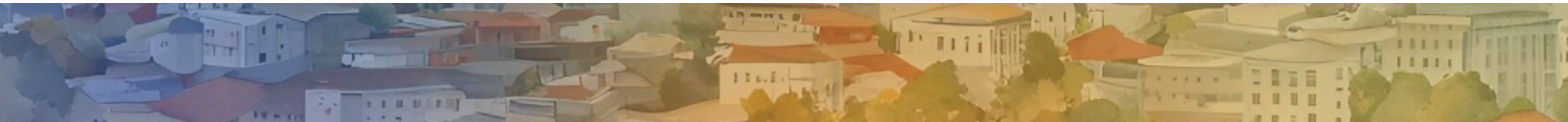
AUTOINFLAMACIÓN VS AUTOINMUNIDAD

- Menos frecuentes
- Mayor peso del componente genético
- Inmunidad innata
- Ausencia de blanco antigénico



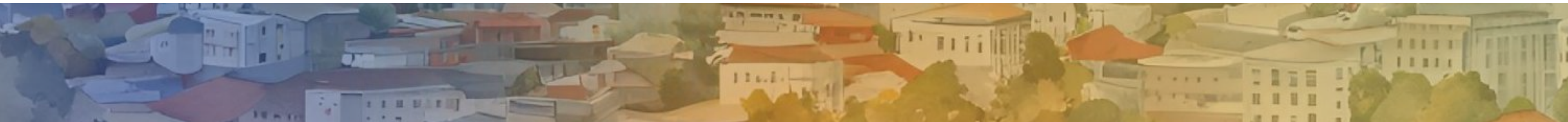
AUTOINFLAMACIÓN VS AUTOINMUNIDAD

- Déficit C1q /déficit C4 □ LES pediátrico (93%/30%)
 - ↑ cutáneo
 - ↓ renal
 - ausencia aDNA; C3 y C4 en rango
 - Tto □ reemplazo C, TPH (C1q def.)
- Defectos en la autofagia (portadoras de EGC-X □ LES-like)
- Interferonopatías □ LES-like
- *DADA2* □ PAN



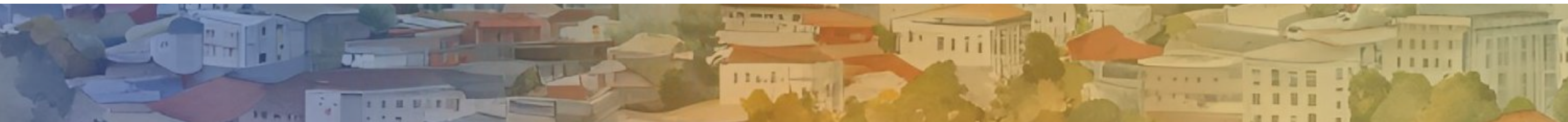
CVID Y AUTOINMUNIDAD

- 20% defecto monogénico
- Influencia poligénica (HLA-DQB1, polimorfismos)
- Complicaciones no infecciosas 30-50% □ LB+, smB-
- Citopenia AI 45%
- Órgano específicas: cutánea, articular, GI, pulmonar



CID

- LOCID y SCID hipomórfico □ 50% AI
- Citopenias, enteropatías, EPI, artropatías, SNC
- *RAG1* (hipo), *ZAP70* (hipo o GOF), *STIM1*
- *WAS* □ AI empeora el pronóstico y es indicación de TPH



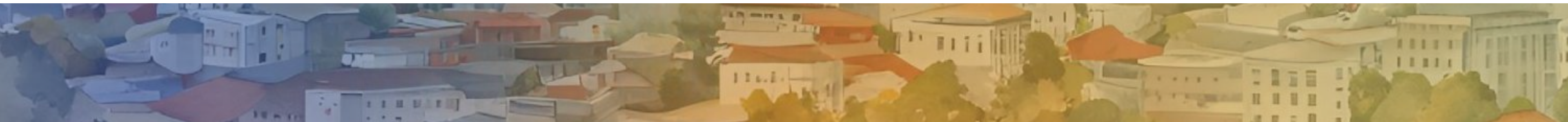
FENOCOPIAS

- Mutaciones somáticas:

- SI □ muy vulnerable a variaciones genéticas
- GOF
- *TLR7, UNC93B1* □ LES
- *UBA1* □ VEXAS

- Autoinmunidad como causa de fenocopia:

- aL17A □ candidiasis crónica mucocutánea
- aIFN γ □ ID con susceptibilidad micobacterias
- aGM-CSF □ proteinosis alveolar
- aC1inh □ AEA
- aCFH □ SHU atipico
- aIFN-I □ Síndrome de Good



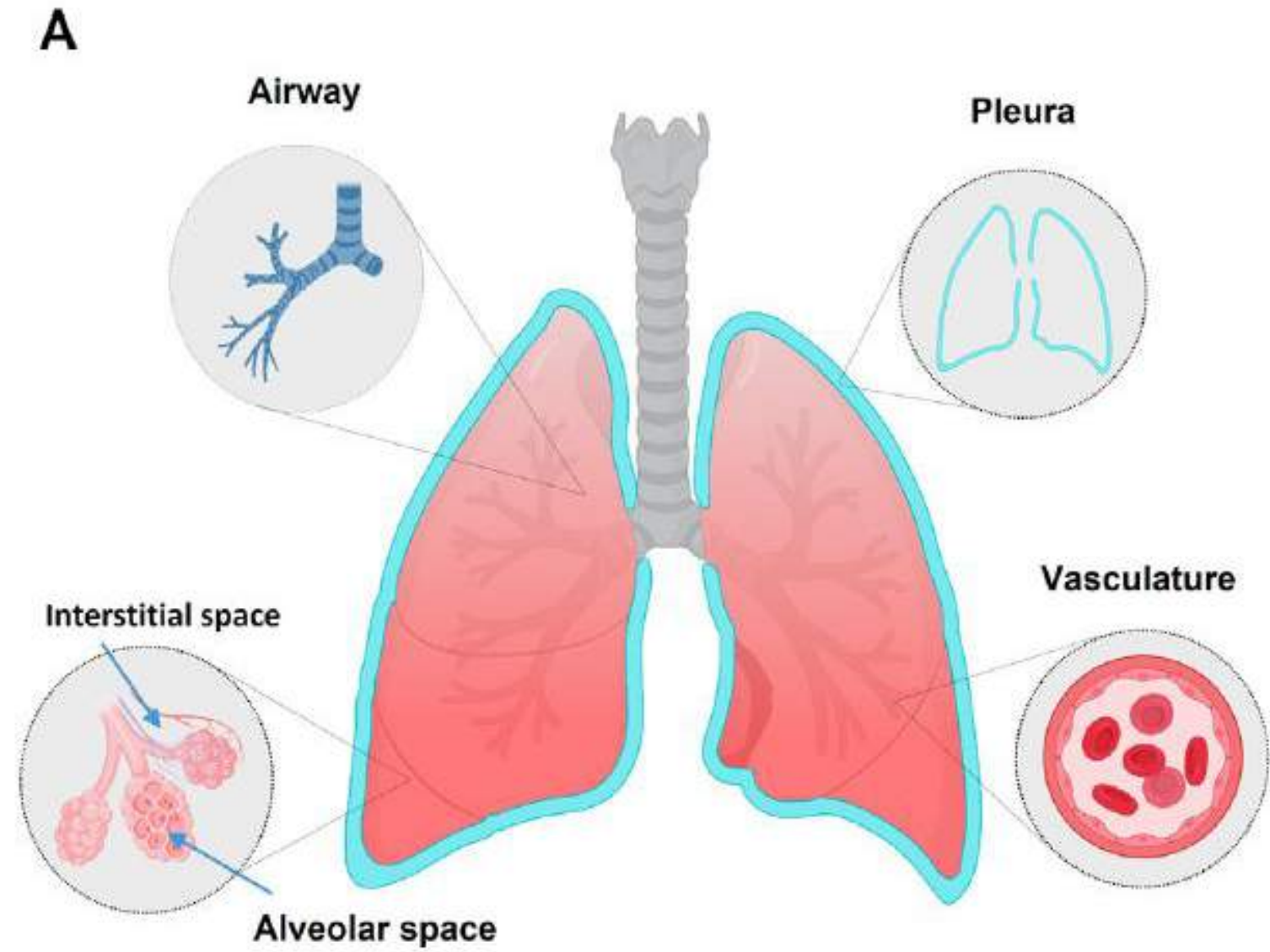


MANIFESTACIONES RESPIRATORIAS

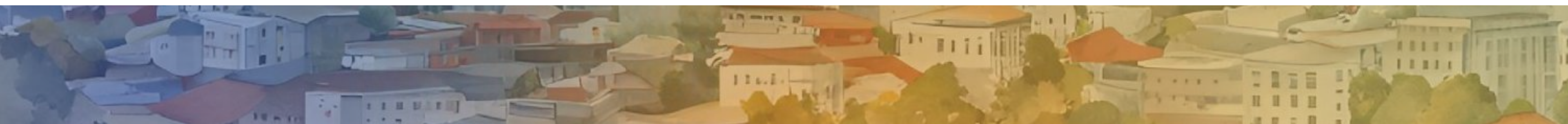
MANIFESTACIONES RESPIRATORIAS

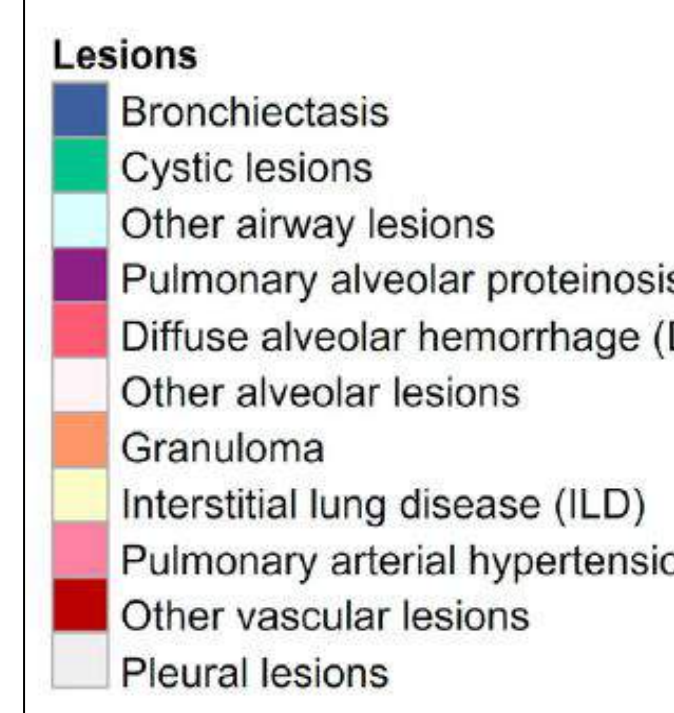
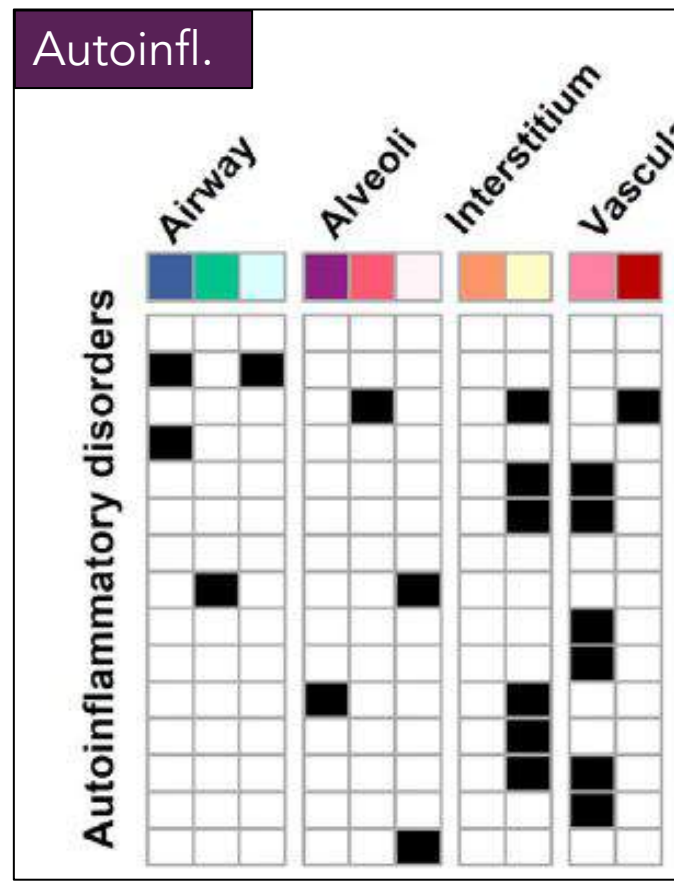
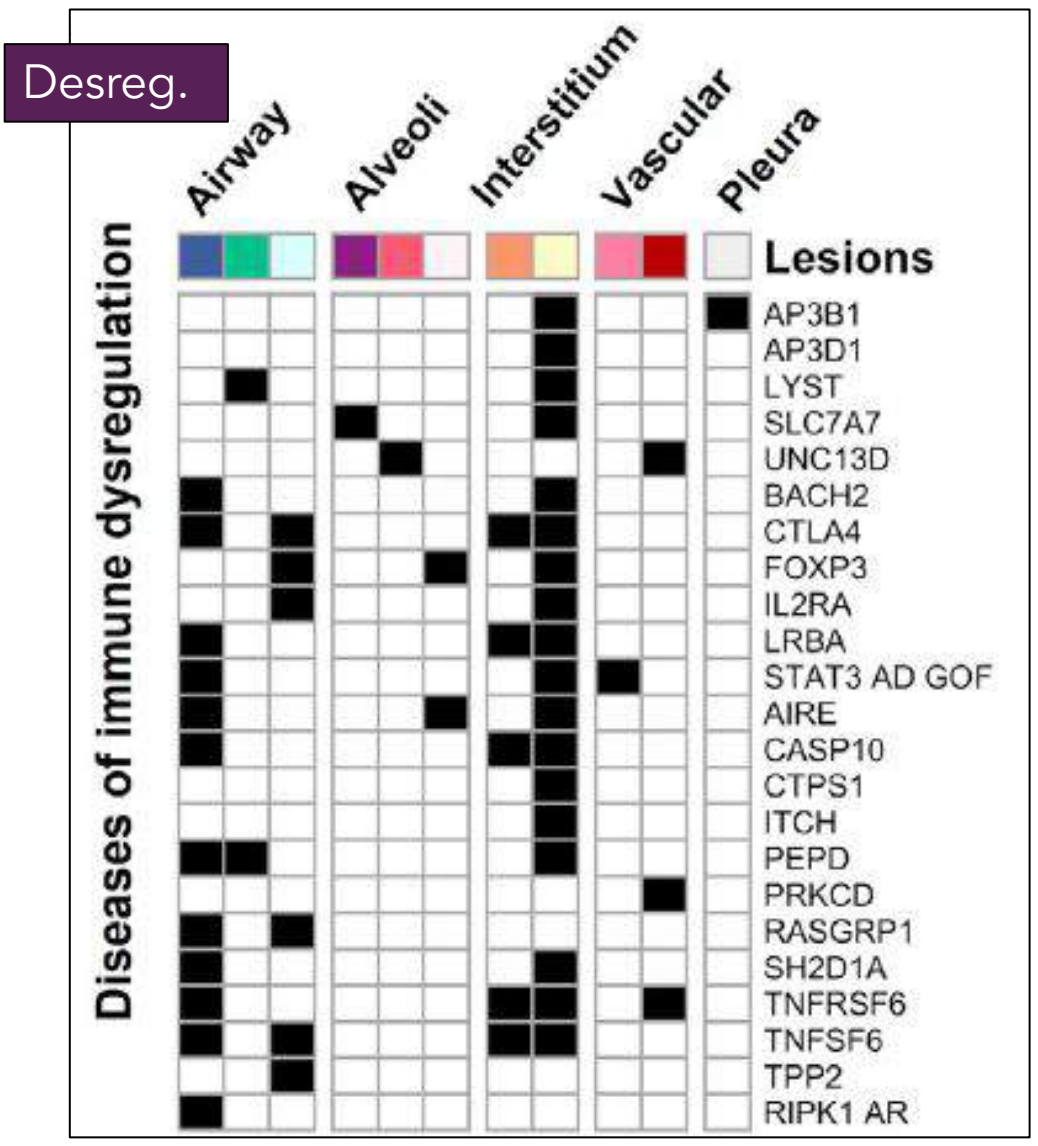
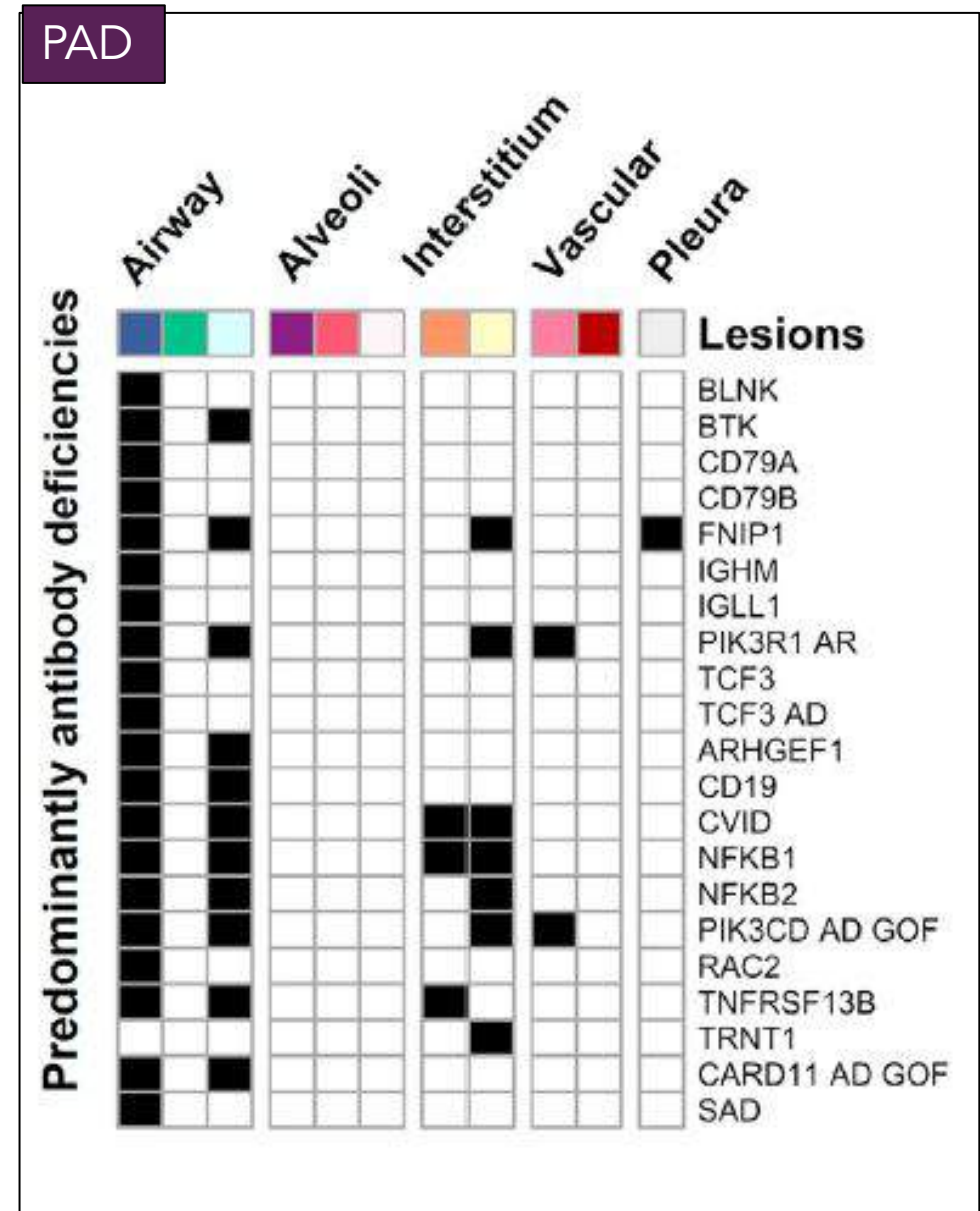
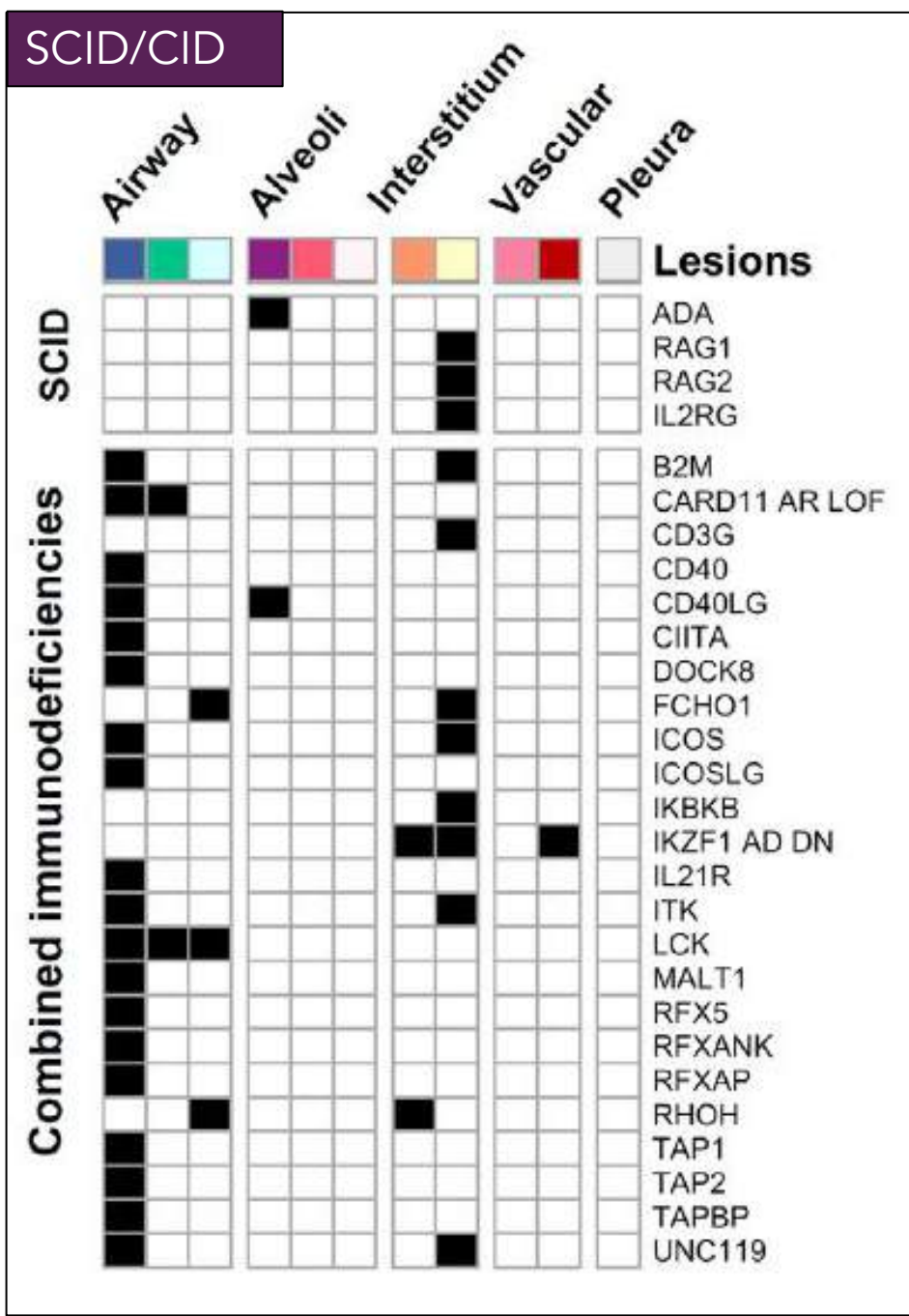
Componente
inmunitario
Interfase

Todas las
categorías de
EII



Sullivan N et al. Curr Opin Allergy Clin Immunol. 2023 Dec 1;23(6):500-506
Gutierrez et al. J Allergy Clin Immunol. 2022 Dec;150(6):1314-1324. doi: 10.1016/j.jaci.2022.08.024





VÍA AÉREA

Bronquiectasias

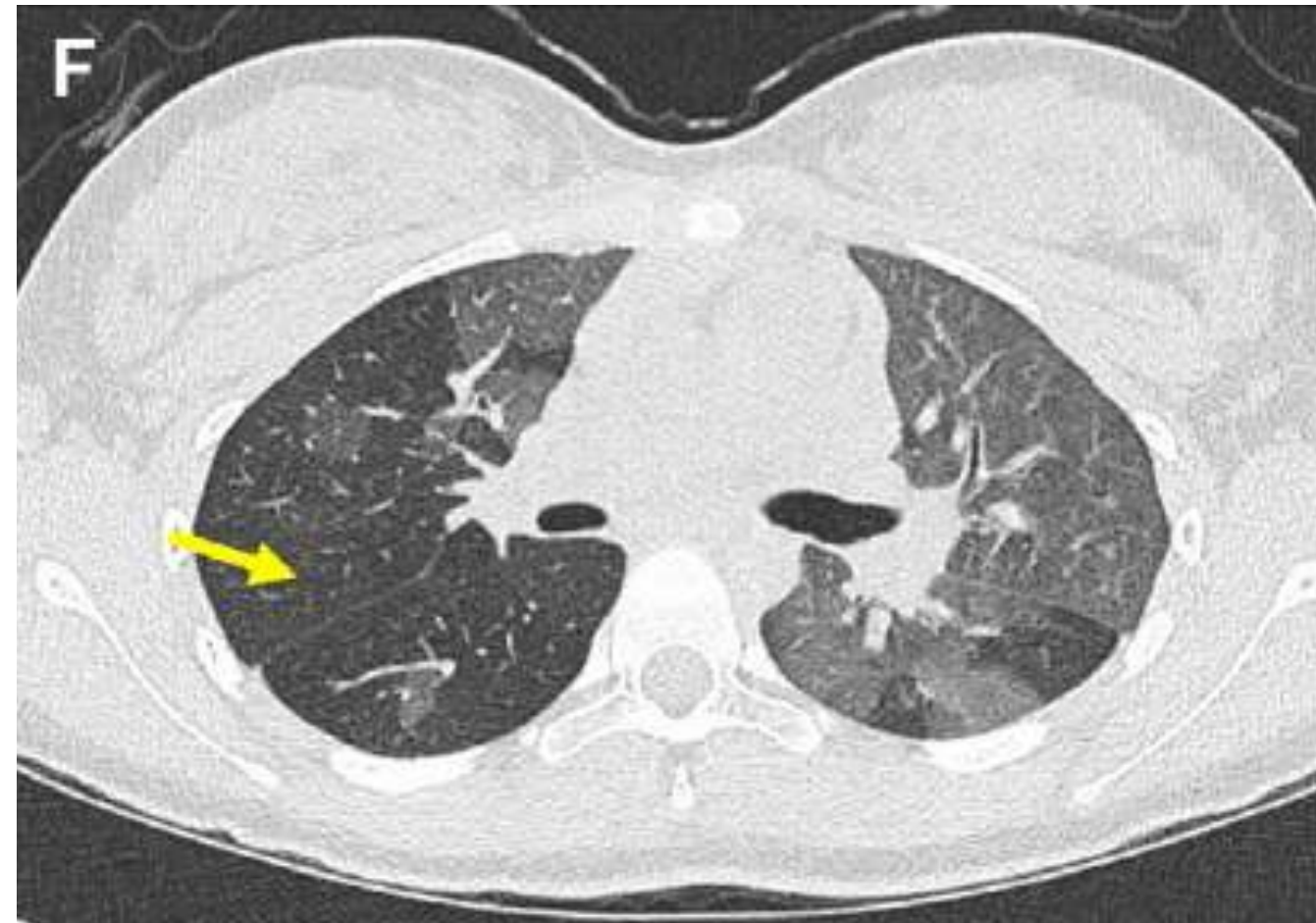
PAD
CID
Fagocitos

Bronquiolitis
obliterante

SCID
AT
CVID

Bronquiolitis
folicular

CVID
CTD

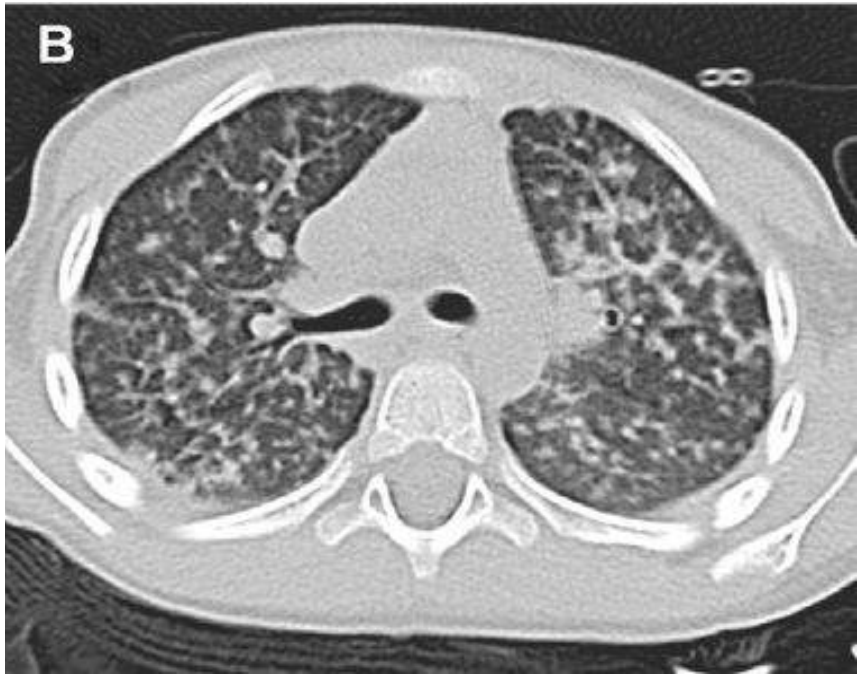
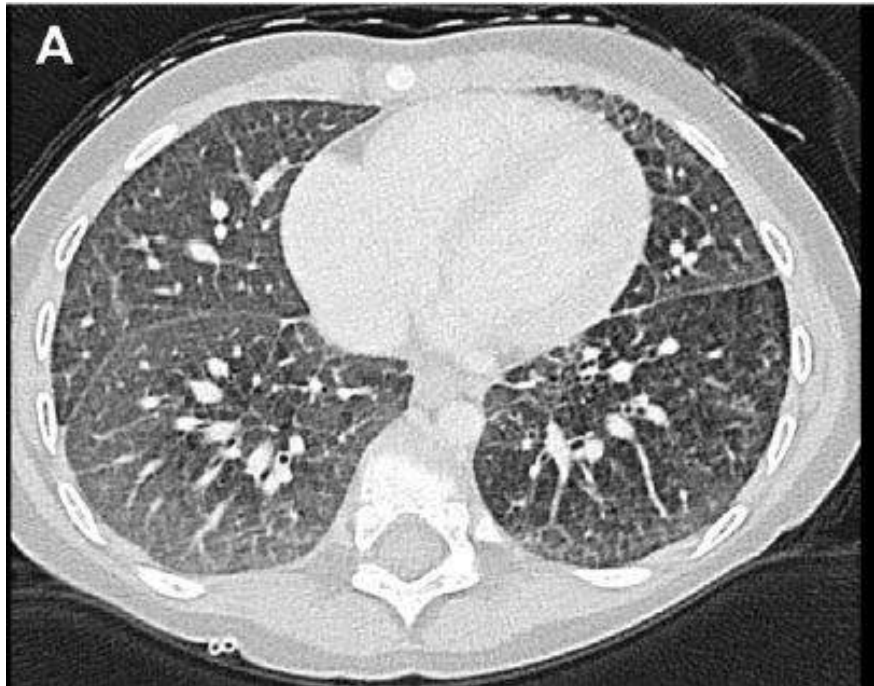


INTERSTICIO

Infiltración
linfocítica

Granulomas

Fibrosis



EPID

LIP

NSIP

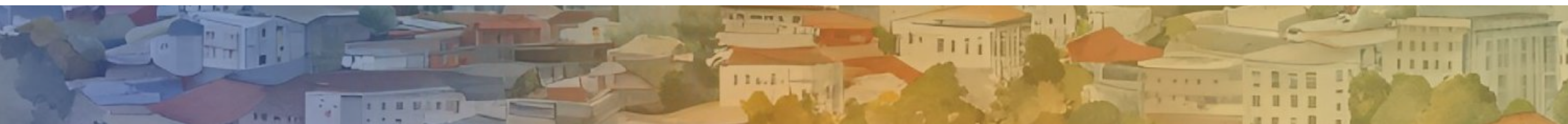
IPF

GLILD

Neumonitis
por HS

Sarcoidosis

Linfoma



ESPACIO ALVEOLAR

Neumonía
organizativa
criptogénica

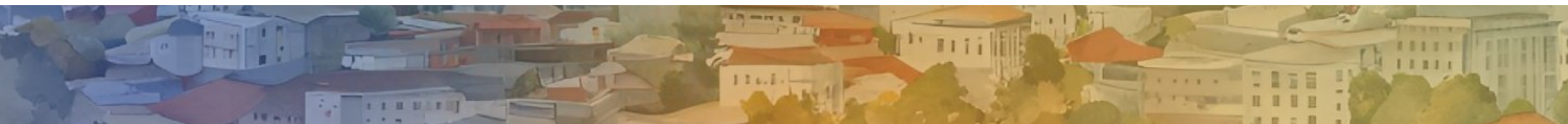
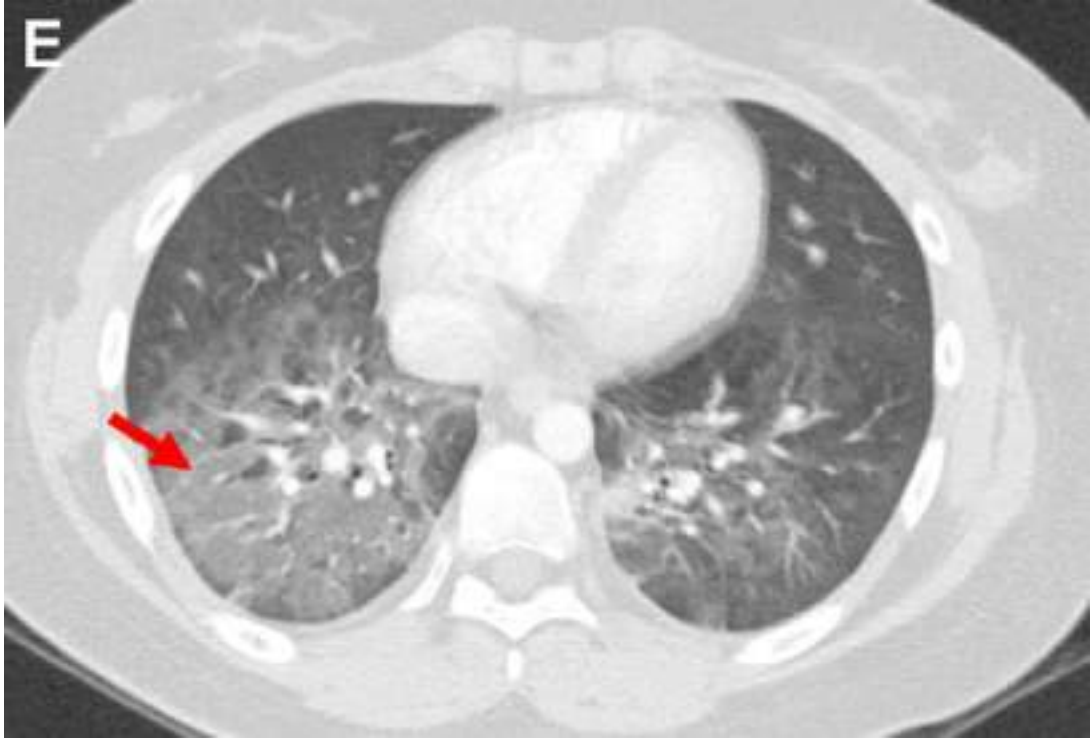
Def. IgM
CVID

Proteinosis
alveolar
primaria

CFS2RA
GATA2
HiperIgM
(CD40L)

Hemorragia
alveolar

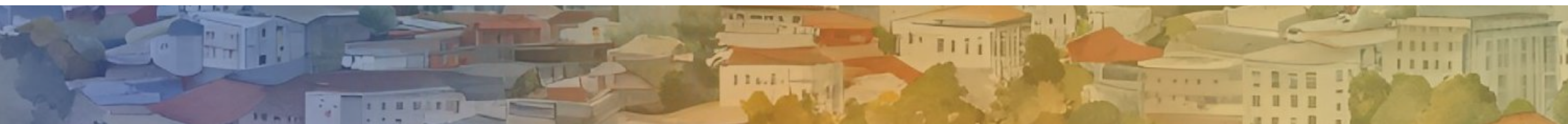
UNC13D
COPA



PLEURA

Pleuritis en contexto de poliserositis

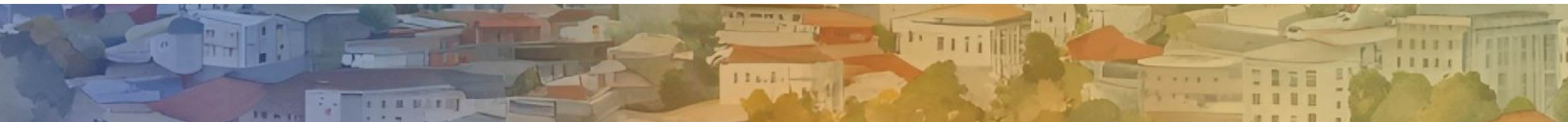
La pleura visceral se conecta con los septos interloblares, puede acompañar patrones de EPI reticular.



VASCULATURA PULMONAR

Hemorragia
alveolar
difusa

Hipertensión
pulmonar



EVALUACIÓN PULMONAR



Clínica

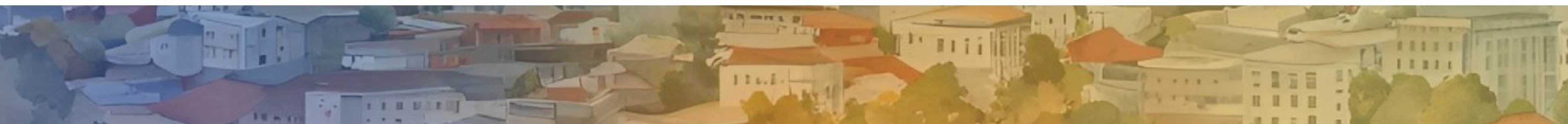
TAC

PFP

Microbiología
AP

Laboratorio
inmunología

Genética



MANEJO MANIFESTACIONES PULMONARES

Profilaxis ATB

Terapia
inhalatoria

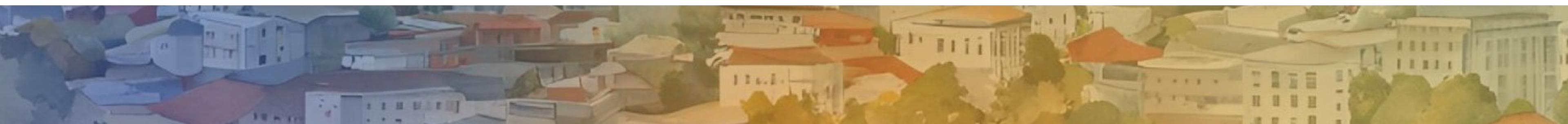
KNT

IgIV
(>1000 mg/dL)

Inmunosupresión

TPH
Tx pulmón

Gutierrez et al. J Allergy Clin Immunol. 2022 Dec;150(6):1314-1324. doi: 10.1016/j.jaci.2022.08.024
Sullivan N et al. Curr Opin Allergy Clin Immunol. 2023 Dec 1;23(6):500-506
Li D et al. J Heart Lung Transplant. 2020 Dec;39(12):1426-1434. doi: 10.1016/j.healun.2020.09.006.
D'Agnano V et al. Pharmaceutics. 2024 Oct 29;16(11):1391. doi: 10.3390/pharmaceutics16111391
Hayoun C et al. Eur Respir J. 2024 Jun 28;63(6):2400518. doi: 10.1183/13993003.00518-2024



INMUNOSUPRESIÓN

Corticoides

- 1ª línea GLILD

Micofenolato

AZA

RTX

- 2ª línea

Terapias dirigidas

- CTLA4 □ Abatacept
- PIK3CD GOF □ leniolisib
- LT CD45O/LT CD45RA □ rapamicina
- GOF STAT3 □ JAKi, tocilizumab

Gutierrez et al. J Allergy Clin Immunol. 2022 Dec;150(6):1314-1324. doi: 10.1016/j.jaci.2022.08.024

Sullivan N et al. Curr Opin Allergy Clin Immunol. 2023 Dec 1;23(6):500-506

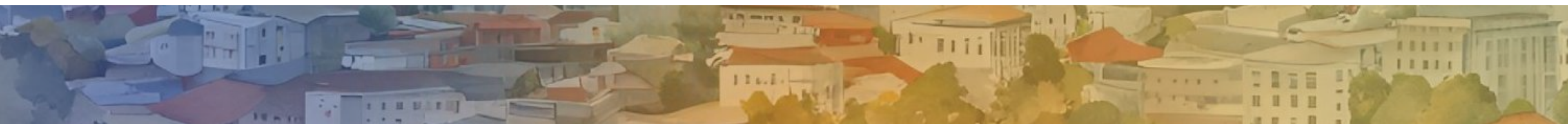
Li D et al. J Heart Lung Transplant. 2020 Dec;39(12):1426-1434. doi: 10.1016/j.healun.2020.09.006.

D'Agnano V et al. Pharmaceutics. 2024 Oct 29;16(11):1391. doi: 10.3390/pharmaceutics16111391

Hayoun C et al. Eur Respir J. 2024 Jun 28;63(6):2400518. doi: 10.1183/13993003.00518-2024

CVID

- Bronquiectasias, bronquiolitis folicular, EPID, HTP (0.4%)
- CVID B+ smB-
- EPID □ 10-20% (LIP, GLILD (8-22%)
- 64% alteraciones en el TAC
- > 50% alteraciones FP
- Esplenomegalia, linfadenopatías y linfopenia T.
- La EPID en CVID eleva la mortalidad (20%)



CONSENSO BRITÁNICO GLILD (2017)

Estudio



- Clínica
- TACAR
- PFP
- FBC
- Bp qx
- Genético

Diagnóstico



- Dg dif.: infección, NO, LIP, sarcoidosis, linfoma
- Confirmación dg: Bp.

Manejo



- Multidisciplinario
- Optimizar IgIV
- Profilaxis ATB
- Iniciar tto: alteración función pulmonar o sintomático con FP normal, pero deteriorándose

CONSENSO BRITÁNICO GLILD (2017)

1ª línea: corticoides sistémicos monoterapia:

1. PDN VO
2. 10 mg/d – 2 mg/kg/d
3. 40 mg (70 kg)

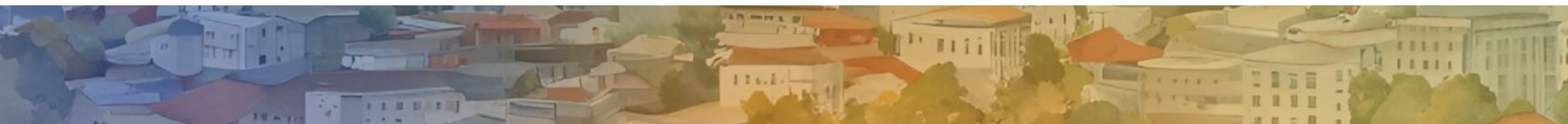
Respuesta a tto: síntomas, PFP (DLCO), TAC.

¿Mantención?

TABLE IV. Consensus on second-line drug therapy in GLILD

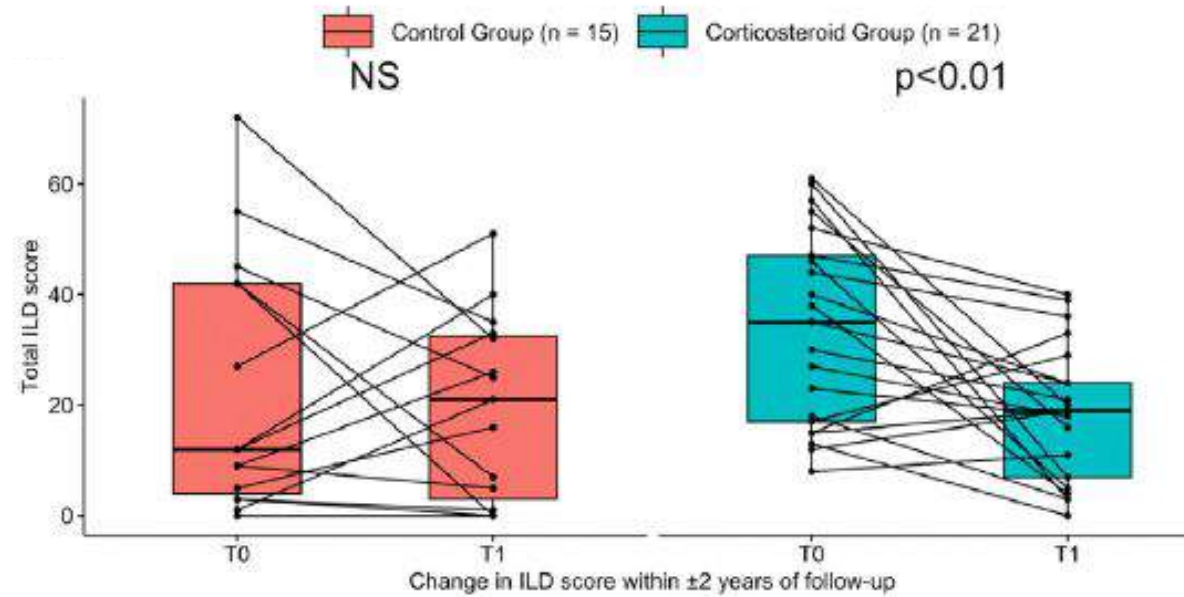
Criteria	No. of respondents	% Agree	% Disagree	Mean ± SD score*
Which of the following drugs would you consider as second-line therapy in GLILD?				
Consensus				
Azathioprine	21	100	0	0.71 ± 0.25
Rituximab	21	90	5	0.67 ± 0.40
Mycophenolate	21	81	5	0.62 ± 0.44
No consensus				
Abatacept ★	18	33	28	0.03 ± 0.50
Anti-TNF agents	17	29	47	-0.12 ± 0.57
Ciclosporin	16	25	25	0.00 ± 0.48
Hydroxychloroquine	19	42	32	0.07 ± 0.56
Methotrexate	17	35	29	0.03 ± 0.51
Sirolimus	18	28	28	0.03 ± 0.53
Tacrolimus	18	22	33	-0.08 ± 0.43

*See text. Scale of -1 (strongly disagree) to +1 (strongly agree), with more extreme scores and smaller SD indicating greater consensus. Consensus defined as ≥80% agreement/disagreement.

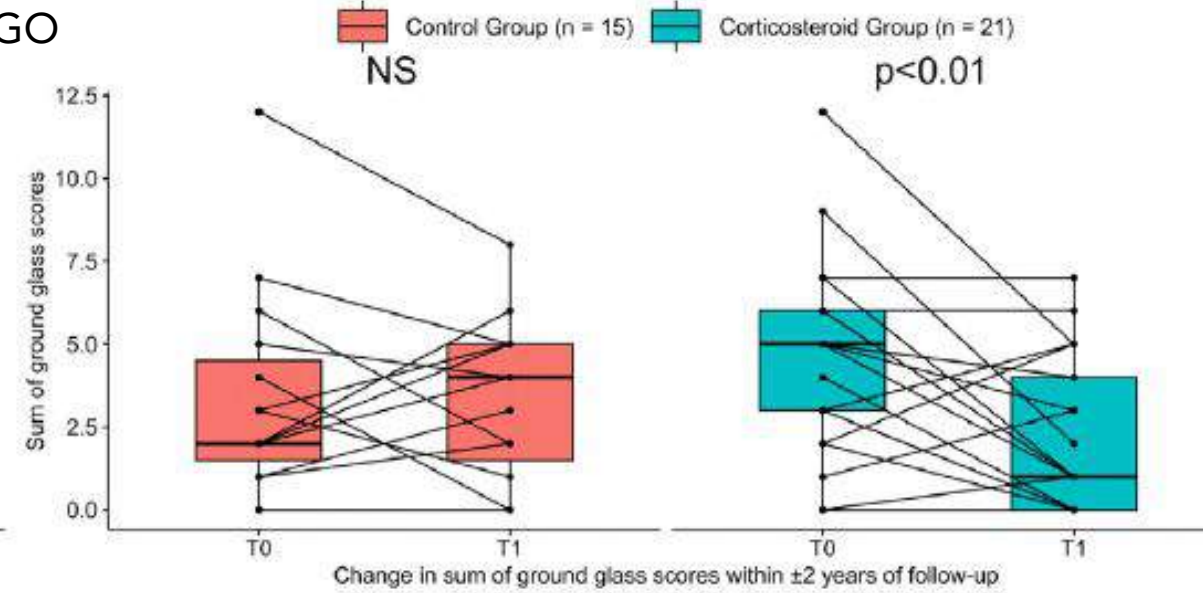


The efficacy and safety of systemic corticosteroids as first line treatment for granulomatous lymphocytic interstitial lung disease

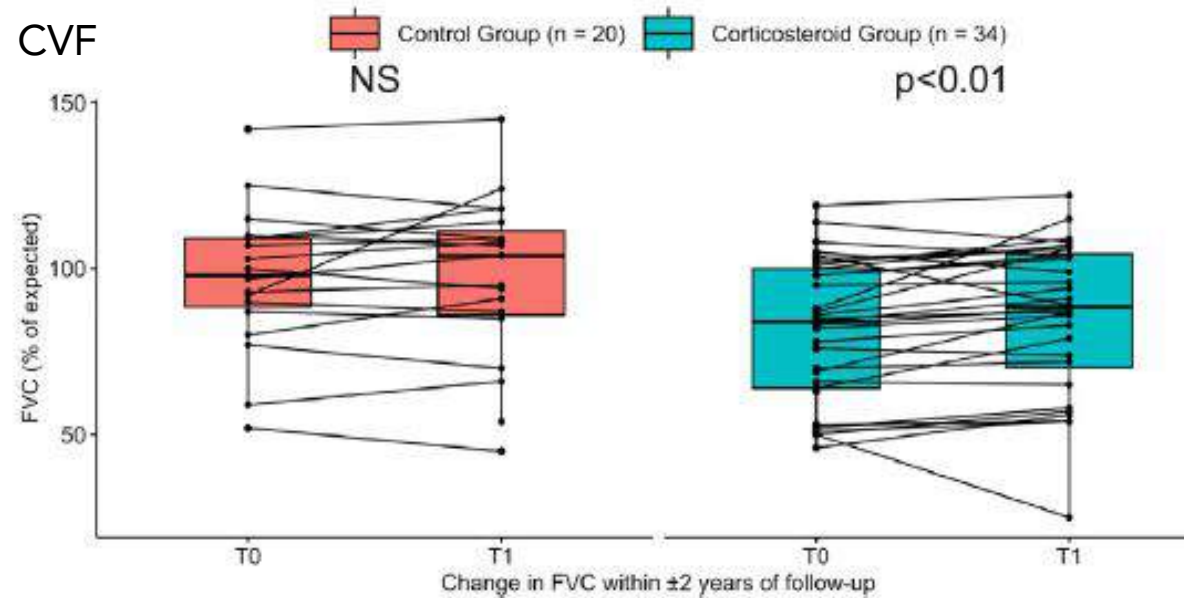
Score ILD



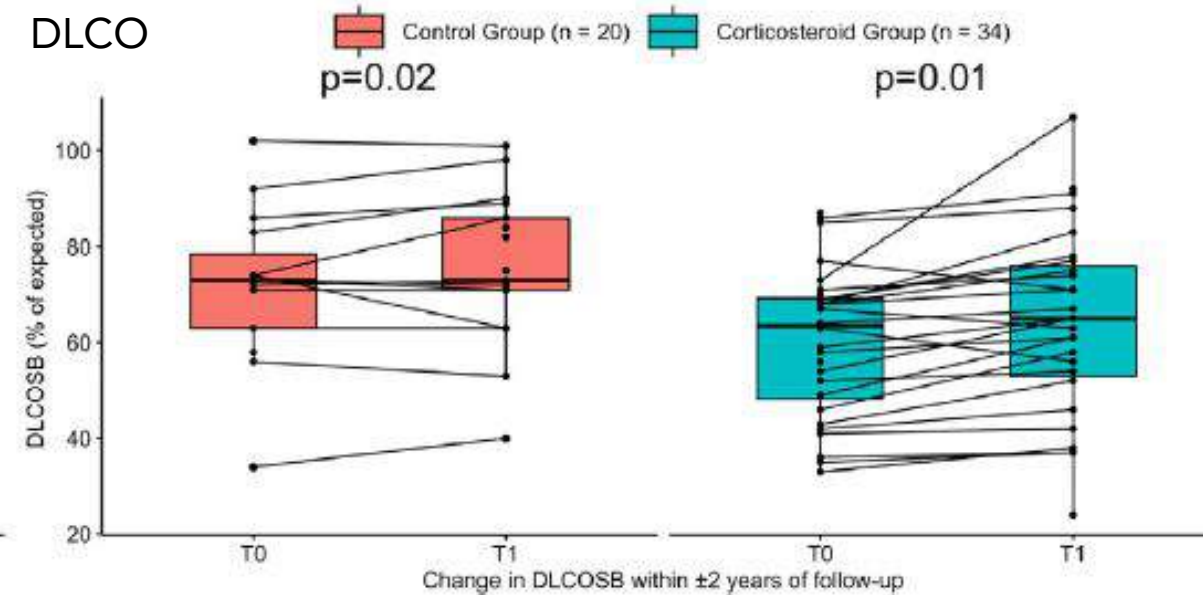
Score GGO



CVF



DLCO



- 56 pacientes (STILPAD)
- EP 1° efectos a los 24 score ILD, score GGO, DLCO
- Dosis media 0.67 mg/día
- Tiempo medio 18 meses
- Respuesta 67%; sostenida (a los 2 años) 42%
- 2 IO (fúngica invasiva complicada); tasa similar a infecciones; OP y Fx
- Mantenimiento □ no pre recaídas

A watercolor illustration of a town with red-roofed buildings and a church spire, set against a backdrop of a large, snow-capped mountain under a blue sky with soft clouds. The scene is rendered in a soft, painterly style with a color palette of blues, greens, and earthy tones.

MANIFESTACIONES DIGESTIVAS

MANIFESTACIONES GASTROINTESTINALES



>50%

INFLAMATORIAS

INFECCIOSAS

AUTOINMUNES

ONCOLÓGICAS

CLINICA SIMILAR

DISTINTA RESPUESTA A
TRATAMIENTO

IDP CON MANIFESTACIONES GASTROINTESTINALES

Research retrieved more than 15 results.
Please precise your search
46 Result found

Thymoma with hypogammaglobulinemia - chronic diarrhea	1/1 7
IL-21 deficiency - chronic diarrhea	1/1 7
NLRC4-MAS (macrophage activating syndrome) - Secretory diarrhea, infantile	1/1 7
NEMO deficiency (IKBKG) - Intractable diarrhea	1/1 7
C5 deficiency - Intractable diarrhea	1/1 7

Proportions of PIDDs by IUIS Category Associated with GI Disease

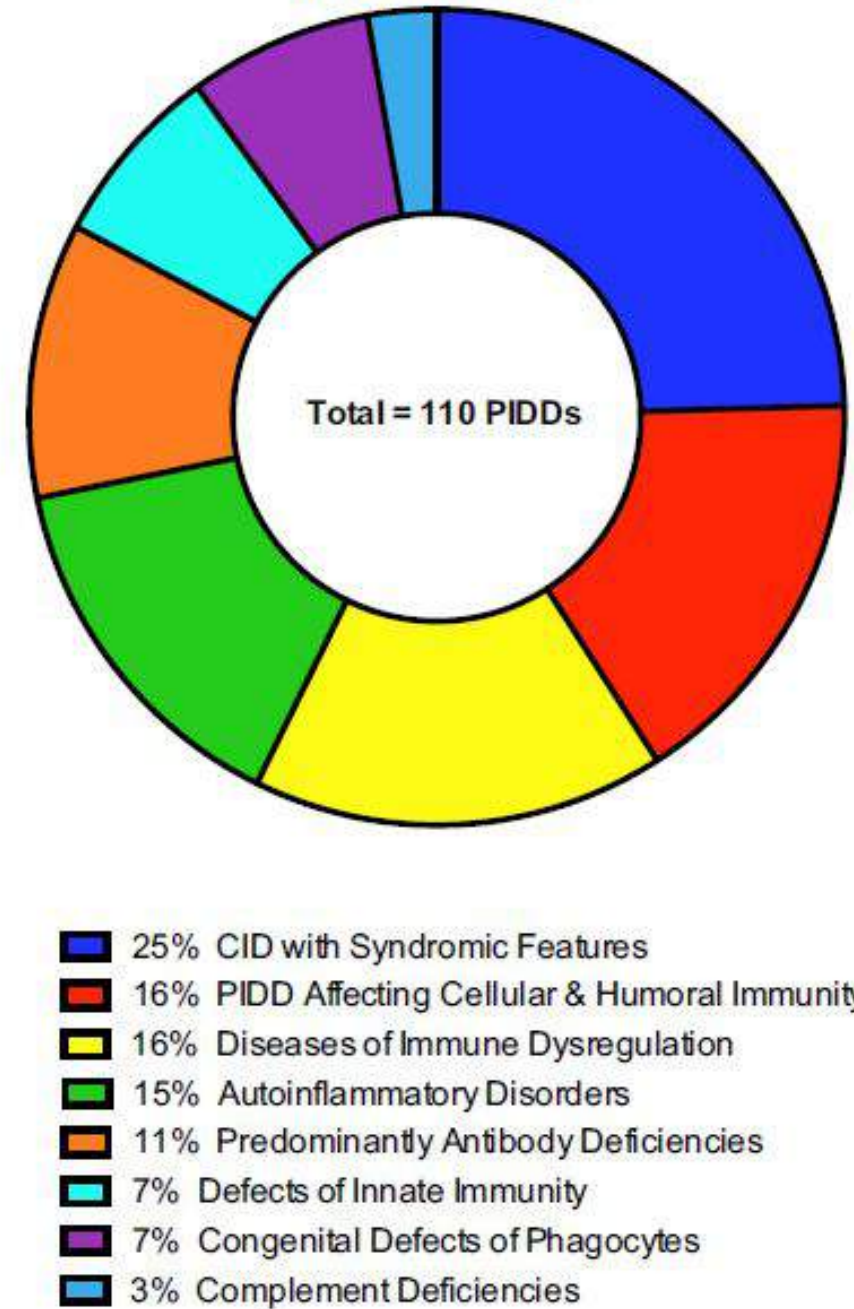


Fig. 1 Proportions of PIDDs by IUIS category associated with GI disease

DIARREA

- 60%
- Malabsorción
- No mejora con IgIV

Infeciosa

Inflamatoria

Tx. motores

Microbiota



Microbiológico



Marcadores inflamatorios



Nutricional



Imagenológico



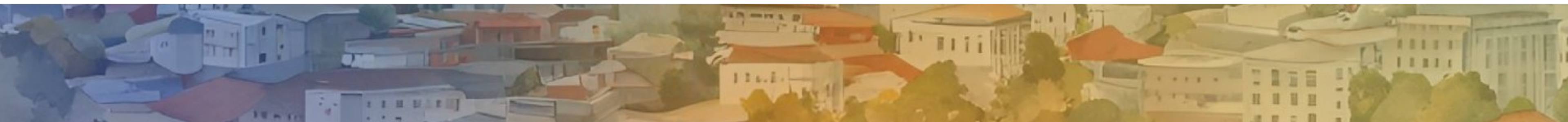
Endoscópico



Histológico

ENFERMEDADES GI EOSINOFÍLICAS

	Number of patients in USIDNET	EoE patients, <i>n</i> (%)	EGID patients, <i>n</i> (%)
Total USIDNET database	5,484	61 (1.1%)	13 (0.2%)
Common variable immunodeficiency	1,820	27 (1.5%)	2 (0.1%)
Chronic Granulomatous Disease	570	4 (0.7%)	2 (0.4%)
Combined immunodeficiency	106	5 (4.7%)	2 (1.9%)
Hyper-IgE Syndrome	104	4 (3.8%)	2 (1.9%)
Autoimmune lymphoproliferative disorder	34	5 (14.7%)	none



CVID

- Diarrea infecciosa (27-50%); SIBO-like (60%)
- No infecciosas □ 15-50%
 - Enteropatía CVID, EC-like (12%), atrofia vellositaria (24-50%)
 - Gastritis AI – anemia perniciosa
 - EII-like (2-13%)
 - Hiperplasia nodular linfoide
 - Enterocolitis colágena
 - Carcinoma gástrico; linfoma
- EDA al dg y cada 24 meses (12 GA, 6 displasia)
- Evaluación nutricional

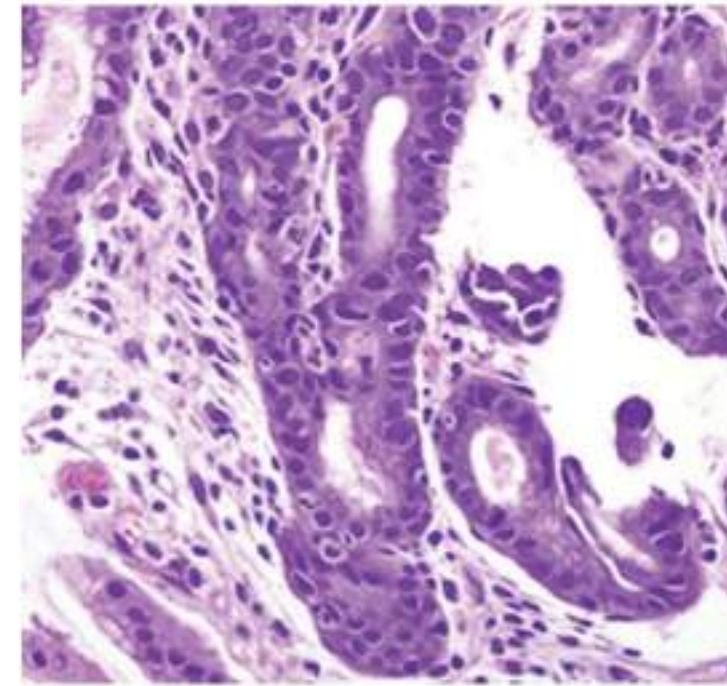


FIGURE 4. There are intraepithelial neutrophils and prominent epithelial apoptosis in this gastric antral biopsy. Despite the active inflammation, note that there are no plasma cells in the lamina propria.

Daniels J et al. *Am J Surg Pathol*. 2007;
Dec;31(12):1800-12. doi:
10.1097/PAS.0b013e3180cab60c

CVID MANEJO

- No hay guías de consenso
- Optimizar IgIV
- ATB (infección, SIBO)
- Nutrición, gluten (HLA)
- Corticoides
- Mesalazina
- Tiopurinas
- aTNF
- Ustekinumab
- Vedolizumab

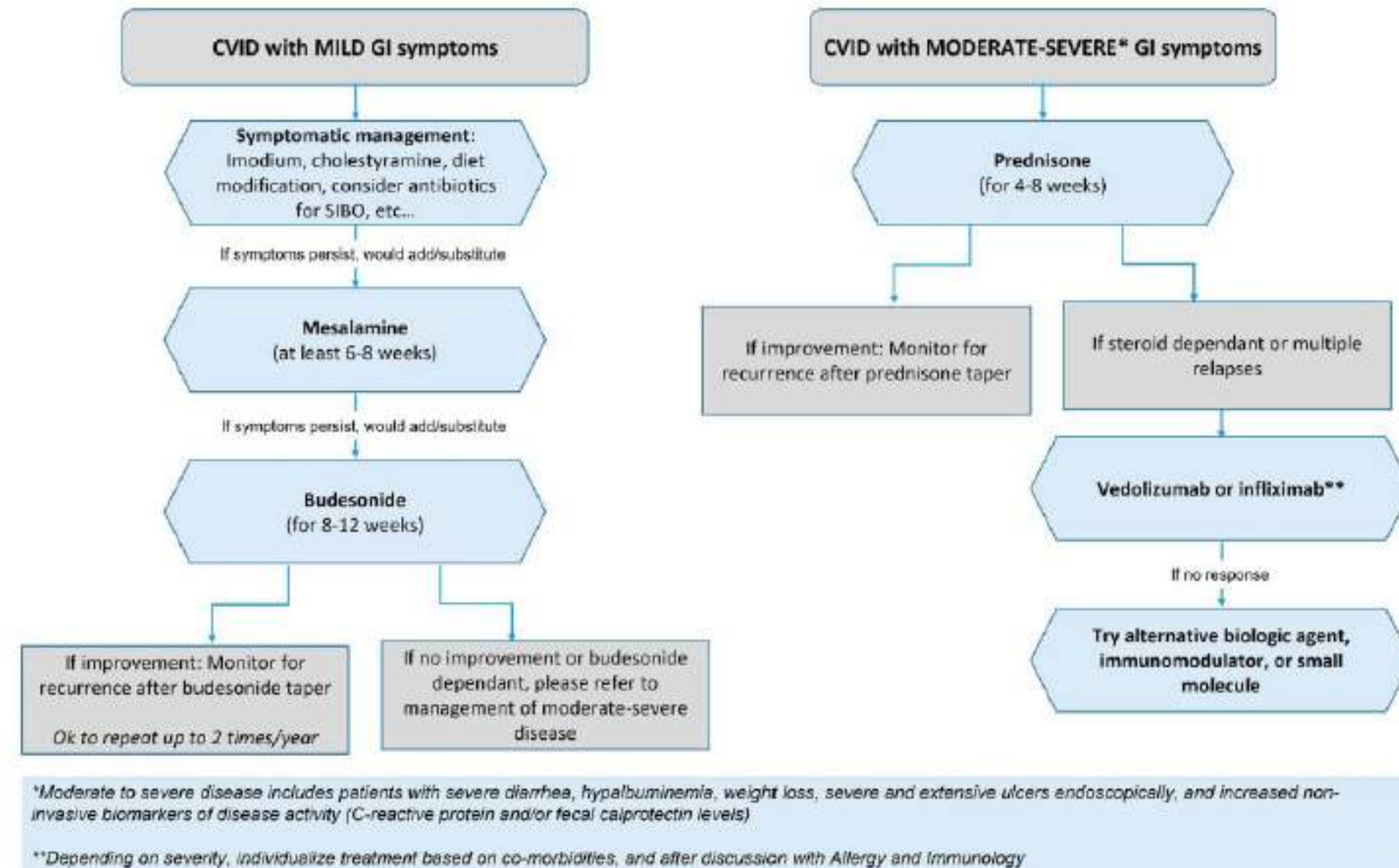
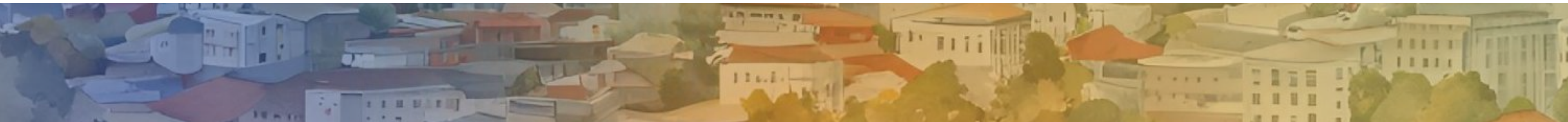


Figure 4. Proposed treatment algorithm for patients with CVID. CVID, common variable immunodeficiency; GI, gastrointestinal; SIBO, small intestinal bacterial overgrowth.

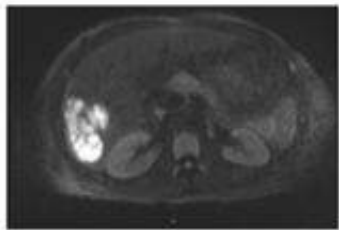
CVID

- Afectación hepática 10%; asociación enteropatía linfocítica
 - Hiperplasia nodular □ HTP no cirrótica, cirrosis
 - Cirrosis biliar primaria
 - Colangitis esclerosante primaria
 - Hepatitis autoinmune
 - Hiperesplenismo □ neutropenia □ infecciones
 - Función hepática, eco/elastografía □ RNM □ biopsia
 - Tx. Hepatico □ SV 55% (3-5 años); infecciones, rechazo, recurrencia



ENFERMEDAD GRANULOMATOSA CRÓNICA

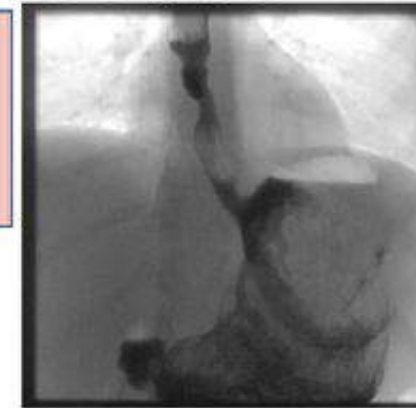
GI/Liver in Chronic Granulomatous Disease



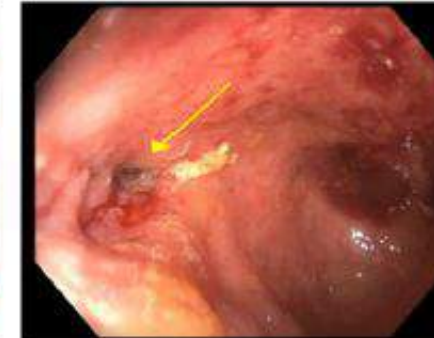
- Liver Abscesses**
- 27-35% of CGD
 - Staphylococcus as leading cause
 - Abx + steroids
- NRH**
- Underrecognized
 - Contributes to morbidity and mortality



- Esophagus and Stomach**
- Dysmotility and strictures
 - Symptoms of dysphagia
 - Dilations and steroids



- Inflammatory bowel disease**
- Up to half of CGD
 - Strictures, fistulas and inflammation
 - Almost always involves the distal colon
 - Surgery, dilations
 - Medical Therapy: Caution with infectious complications



Take Home Points:

- Think of CGD in early or atypical IBD
- IBD is common in CGD
- Liver abscesses may require steroids for complete resolution
- Think of NRH and portal hypertension
- Immunomodulation can result in significant infections
- Multidisciplinary care

A watercolor illustration of a town with a large mountain in the background. The town features buildings with red roofs and a prominent church with a tall spire. The mountain is covered in snow and has a blueish tint. The sky is light blue with soft clouds. A blue rounded rectangle is overlaid on the image, containing the text.

CÁNCER Y LINFOPROLIFERACIÓN

EII Y CÁNCER

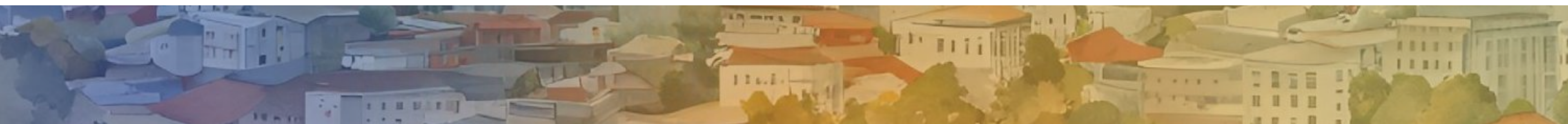
1/3 genes IUIS
rol directo en
oncogénesis

2^a causa de
muerte (<
infecciones)



Mayor
incidencia, más
precoces

Hematológicos
(LNH) y GI

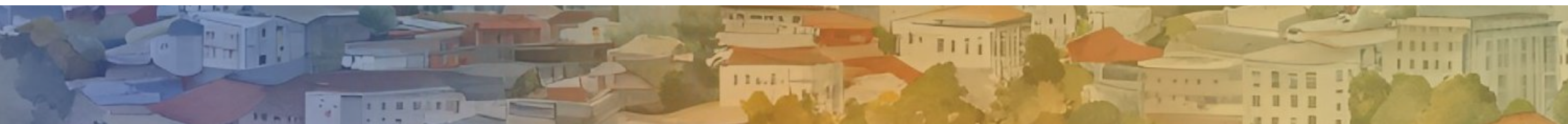
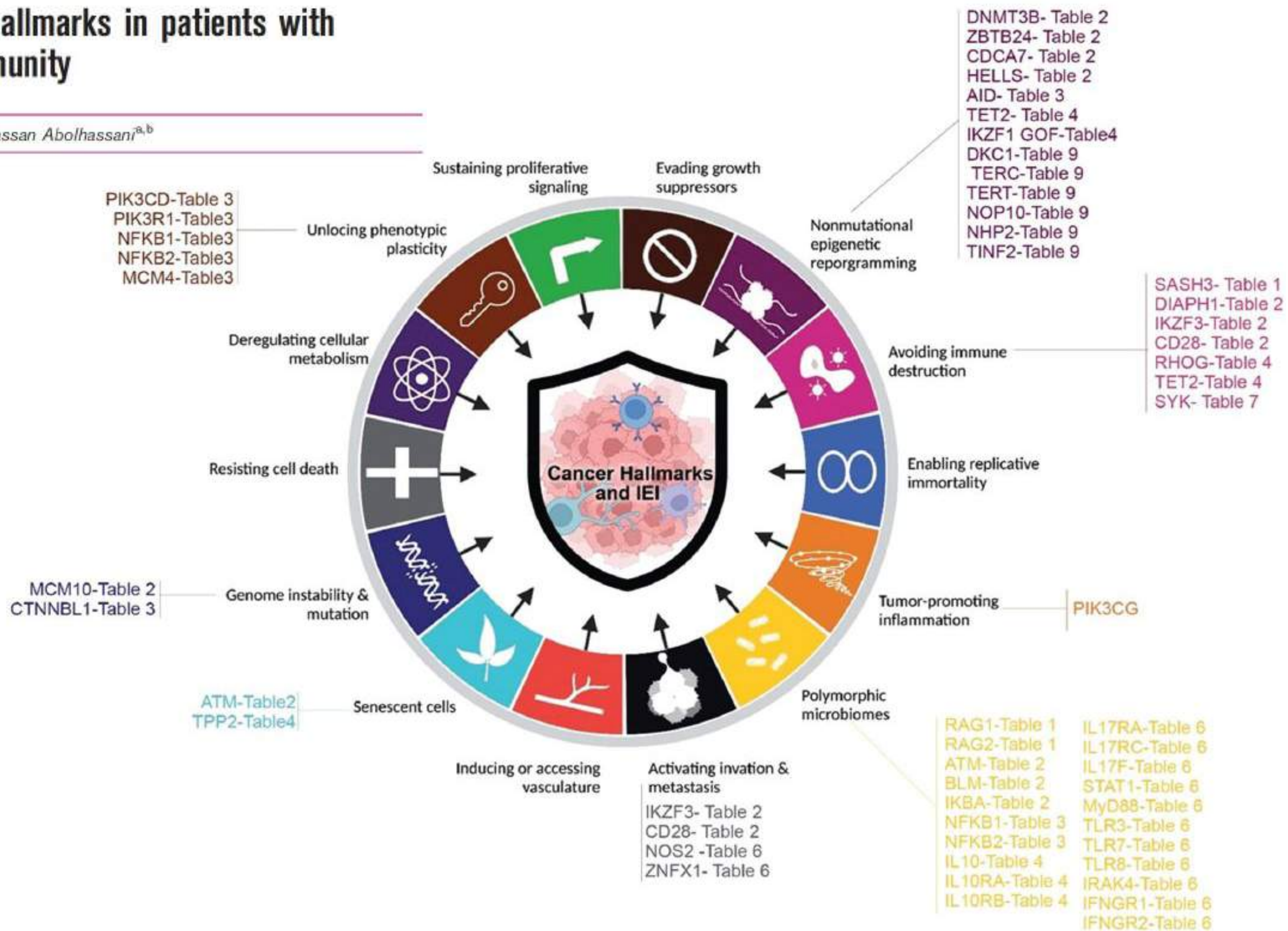


ONCOGÉNESIS EN EII



Updates of cancer hallmarks in patients with inborn errors of immunity

Yating Wang^a and Hassan Abolhassani^{a,b}



Cancer Trends in Inborn Errors of Immunity: A Systematic Review and Meta-Analysis

Saba Fekrvand^{1,2} · Hassan Abolhassani^{1,3} · Zahra Hamidi Esfahani^{1,2} · Najmeh Nameh Goshay Fard⁴ · Mahboube Amiri¹ · Helia Salehi¹ · Amir Almasi-Hashiani⁵ · Ali Saeedi-Boroujeni⁶ · Nazanin Fathi^{1,2} · Maryam Mohtashami^{1,7} · Azadehsadat Razavi^{1,8} · Arash Heidari^{1,2} · Gholamreza Azizi^{9,10} · Shaghayegh Khanmohammadi^{1,2} · Milad Ahangarzadeh¹ · Kiarash Saleki^{11,12,13} · Gholamreza Hassanpour¹⁴ · Nima Rezaei^{1,2} · Reza Yazdani^{1,2}

4607 pacientes

Todos los grupos IUIS
(salvo VIII)

BMF (27%)
CID-Synd. (21%)
Dysreg. (20%)

NBS1 (51%)
XLP (30%)
Fanconi (27%)
AT (16%)

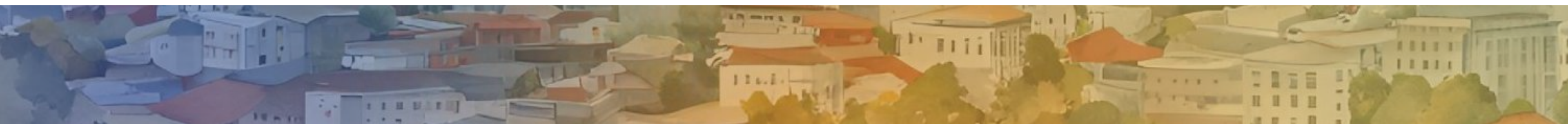
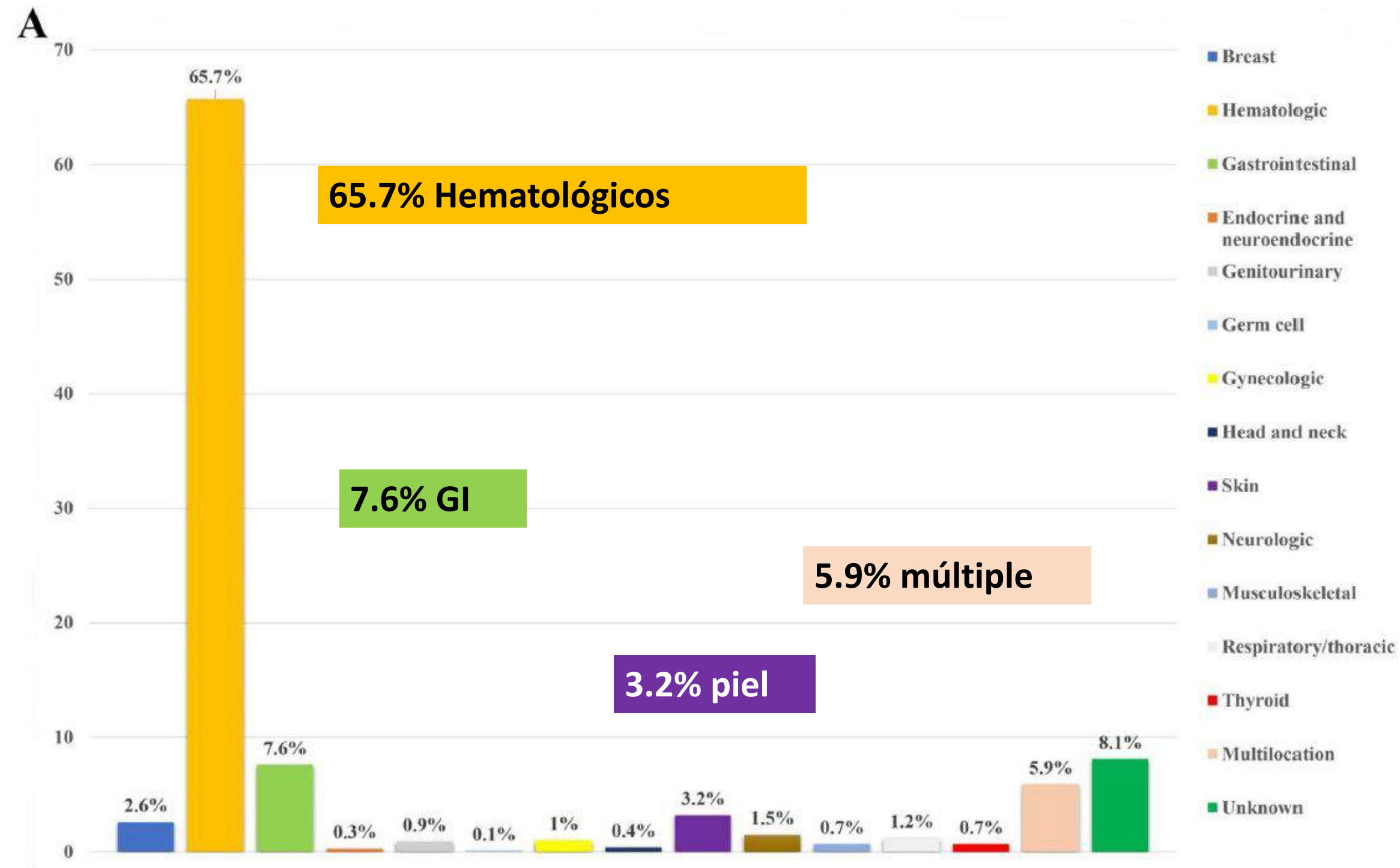
 12%

Dg: 22 años



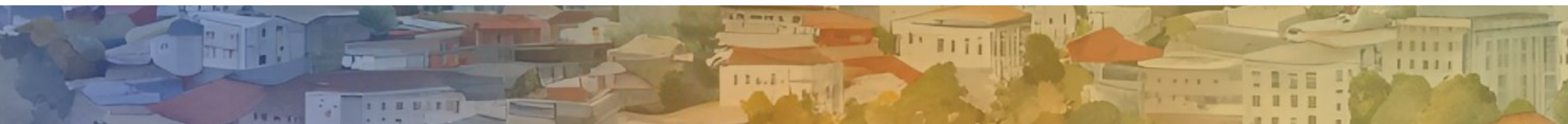
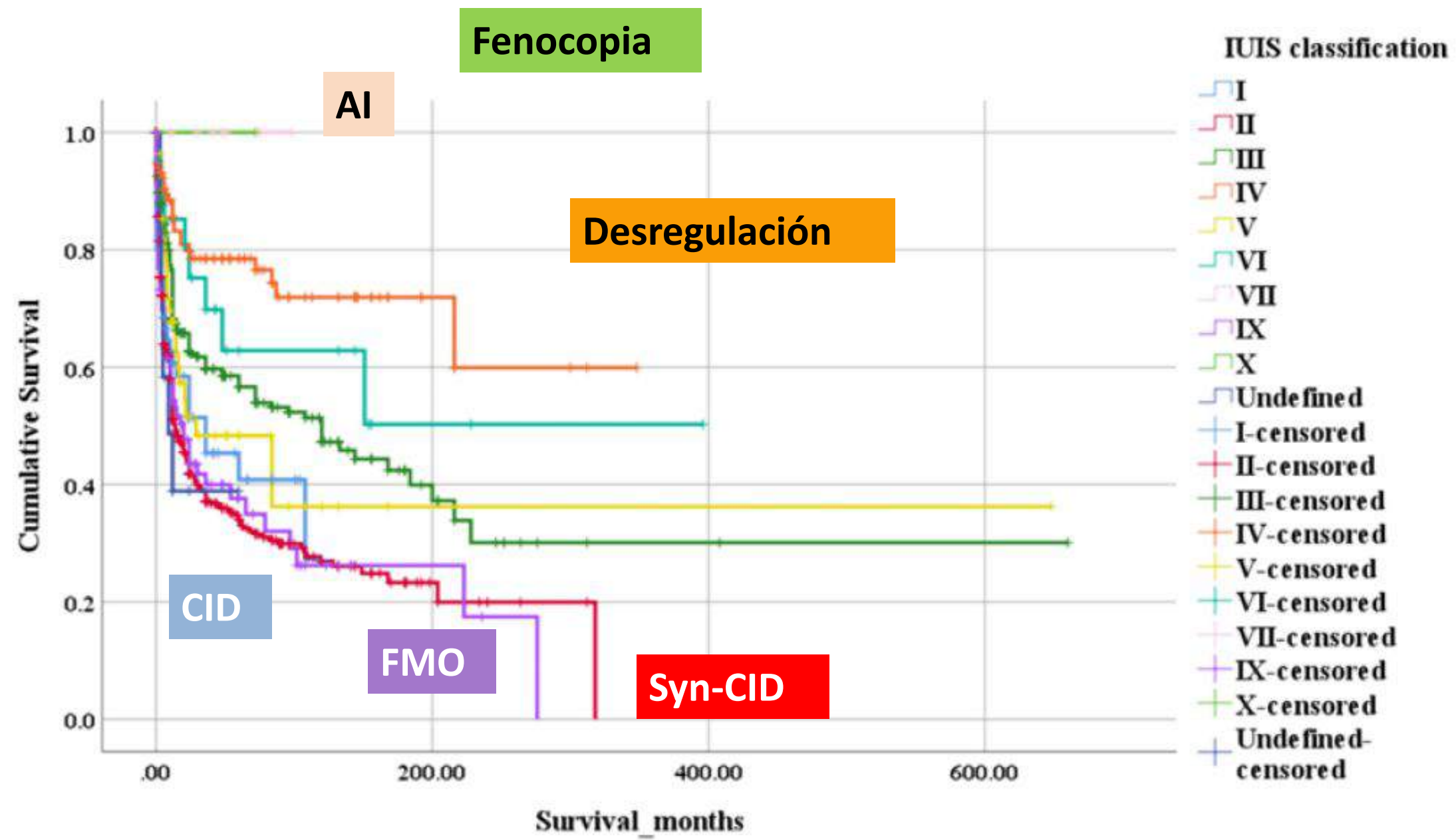
1.2 : 1

Distribución de los tipos de cáncer en pacientes con EII



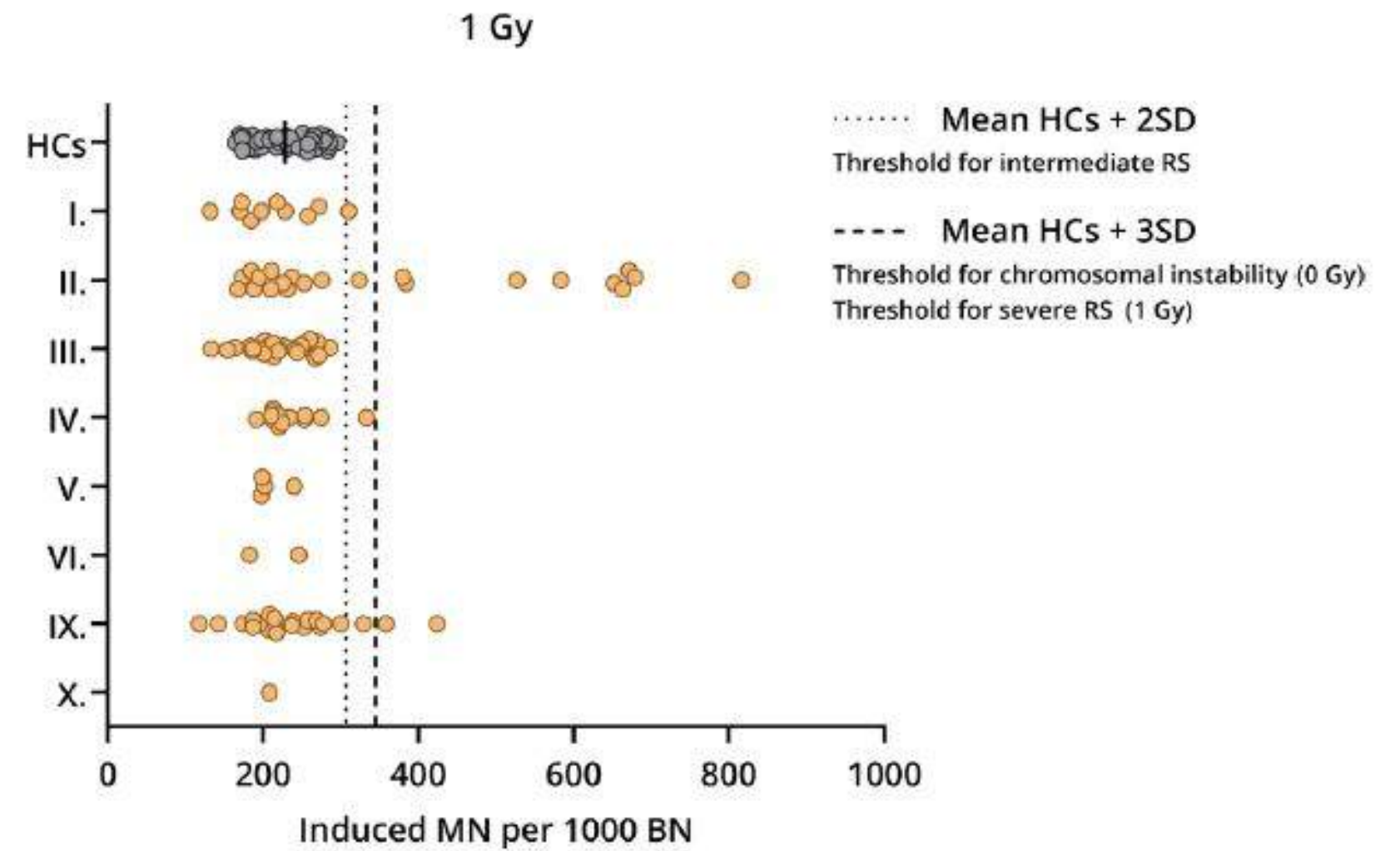
MORTALIDAD Y SV

† 52%



RADIOSENSIBILIDAD

- Ell por defectos en los sistemas de reparación de DNA
- CID/SCID □ Artemis, DNA ligasa IV, ADA, CD40LG
- CID sindrómico □ ATM, NBS1
- CVID
- Desregulación □ LRBA
- FMO □ FANCI, ERCC6L2



LINFOPROLIFERACIÓN

Proliferación policlonal, oligoclonal o monoclonal persistente de células linfoides

Desregulación

Incidencia 0.7-18%
Edad más temprana

Linfoadenopatias
Esplenomegalia
Infiltración extranodal
Linfocitosis SP



ALPS

XLP

CVID

APDS

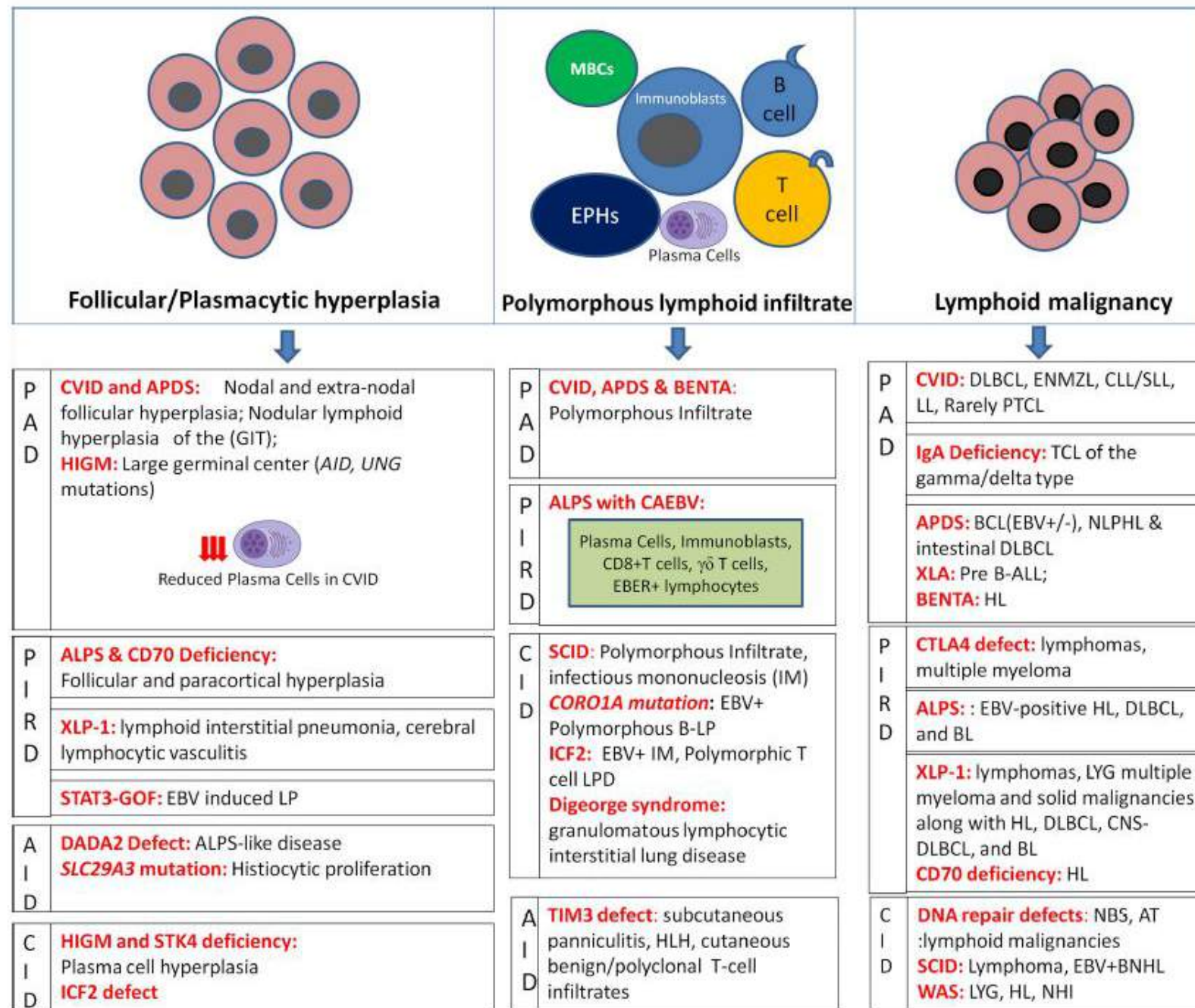
VEB-r



50%



HISTOPATOLOGÍA SLP



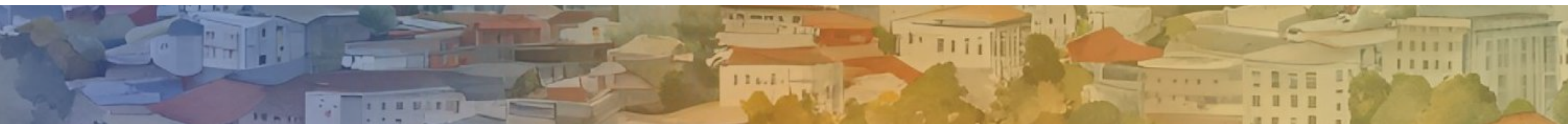
Hiperplasia linfoide células B

LP polimorfa LB

Linfoma indolente

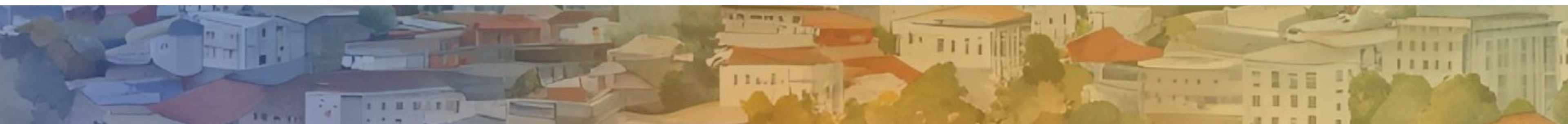
LNH agresivo

LH



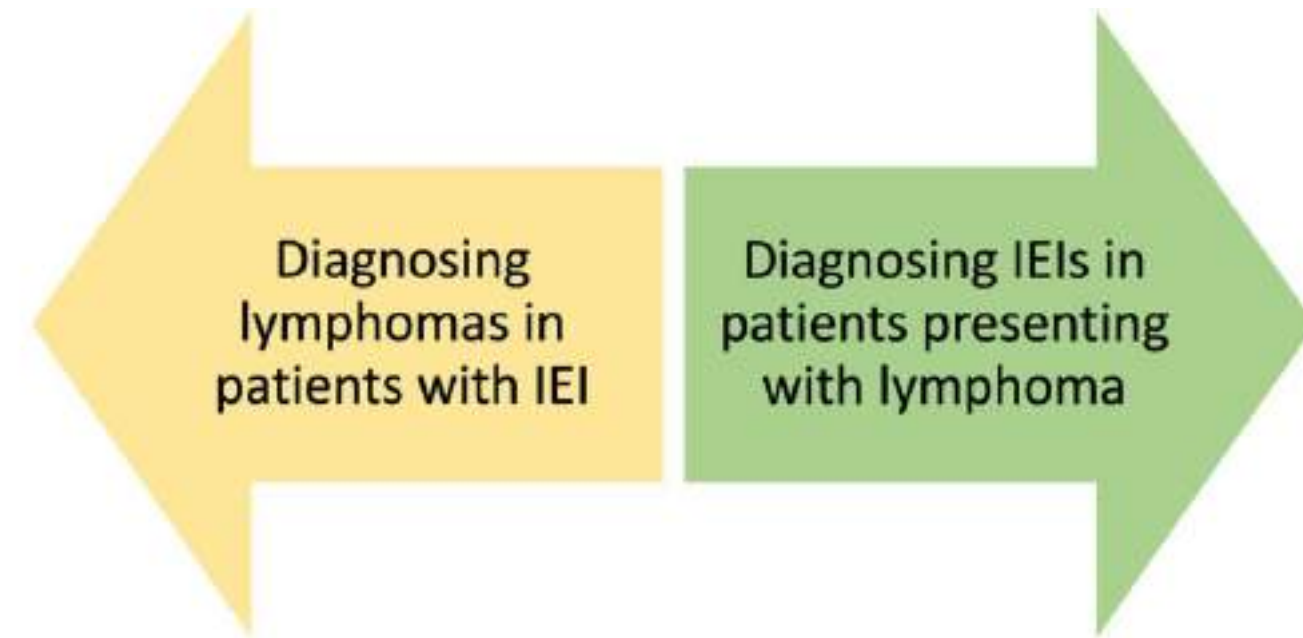
CVID-LP

- 20% LP
- LB Tr hi; TACI, NFKB1
- Linfoma 8% (USIDNET); 2ª causa de mortalidad
- Linfoma difuso CB grandes, linfoma extranodal de la zona marginal, LH
- LP benigna: hiperplasia folicular nodal y extranodal, hiperplasia nodular linfoide GI clonal no maligna (sin células plasmáticas)
- Proliferación LTCD8 (SP, hígado) □ diferencia leucemia linfocítica granular de LT grandes



ENFRENTAMIENTO

- Importancia diagnóstico genético
- Evaluación periódica
- Tratamiento manifestaciones autoinmunes, autoinflamatorias, infecciosas
- Prevención exposiciones
- Linfoma B: QT + aCD20 □ TPH alogénico con acondicionamiento intensidad reducida
- VEB (+) pre y post TPH □ tratar (RTX, EBV-CTLs)
- SLP no neoplásico □ RTX, mTOR (sirolimus, rapamicina)





SALUD CARDIOVASCULAR

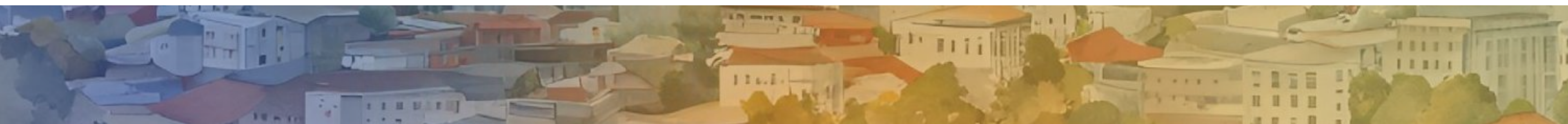
ENFERMEDAD CARDIOVASCULAR

- Datos de pacientes ID secundaria, ETC
- FRCV convencionales + propios

Autoinmunidad

Infecciones

HipolgG



Higher Cardiovascular Risk in Common Variable Immunodeficiency and X-Linked Agammaglobulinaemia Patients

Daniele Gonçalves Vieira^{a, b} Beatriz Tavares Costa-Carvalho^a Sonia Hix^c
Rosângela da Silva^d Milena S.G. Correia^c Roseli Oselka Saccardo Sarni^{a, c}

^aFederal University of Sao Paulo, Sao Paulo, Sao Paulo, ^bMidwest State University, Guarapuava, Paraná, ^cABC University, Santo Andre, Sao Paulo, ^dFederal University of Alfenas, Alfenas, Minas Gerais, Brazil

Table 1. Demographic and anthropometric data in CVID and XLA patients and healthy controls

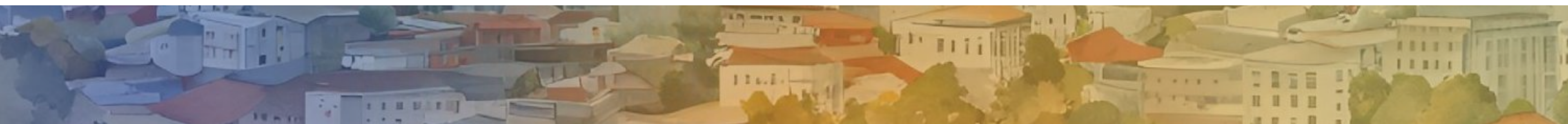
Variables	Patients (n = 24)	Controls (n = 12)	p value
Age, years	32.08±13.66	29.33±13.74	0.576 [†]
Gender			
Male, %	15 (62.5)	6 (50)	0.473 [‡]
BMI, kg/m ²	22.5±5.1	21.3±4.0	0.448 [†]
Underweight, %	3 (12.5)	1 (8.3)	
Normal, %	13 (54.2)	10 (83.3)	0.203 [‡]
Overweight, %	8 (33.3)	1 (8.3)	
WC, cm	81.1±13.0	78.5±14.3	0.604 [†]
Adequate, %	15 (62.5)	9 (75)	0.453 [‡]
High, %	9 (37.5)	3 (25)	
Body fat, %	26.4±9.9	24.1±8.2	0.477 [†]
Adequate, %	7 (29.2)	5 (41.7)	0.396 [‡]
High, %	17 (70.8)	7 (58.3)	

[†] t test; [‡] χ^2 -test.

Table 2. Lipid profile, apo A-I, MPO, CETP, LCAT, CRP, and TNF-alpha in CVID and XLA patients and healthy controls

	Patients (n = 24)	Controls (n = 12)	p value
TC, mg/dl	162±34	185±32	0.078 [†]
HDL-c, mg/dl	43±18	57.5±16	0.025 [†]
NHDL-c, mg/dl	119±42	127±23	0.471 [†]
LDL-c, mg/dl	96±25	109±24	0.178 [†]
TG, mg/dl	83 (46–530)	65 (38–236)	0.455 [§]
sdLDL, mg/dl	20 (8–106)	24 (9–46)	0.455 [§]
apo A-I, mg/dl	137±17	161±26	0.013 [†]
MPO, µg/l	273 (70–1,220)	220 (104–917)	0.986 [§]
CETP, µg/ml	1.4 (1–7)	1.4 (0.7–2.5)	0.614 [§]
LCAT, µg/ml	4.6±1.12	6.1±2.3	0.052 [†]
CRP, mg/dl	1.6 (0.02–13.4)	0.45 (0.05–1.2)	0.008 [§]
TNF-alpha, pg/ml	10.5 (4.77–32.0)	2.35 (0–6.5)	<0.001 [§]

[†] t test; [§] Mann-Whitney test.



Common variable immunodeficiency—an independent risk factor for atherosclerotic cardiovascular diseases

Juho Mattila^{1*}, Niina Pitkänen² and Hannu Järveläinen^{1,3}

¹Institute of Biomedicine, University of Turku, Turku, Finland, ²Auria Biobank, Turku University Hospital and University of Turku, Turku, Finland, ³Department of Internal Medicine, Satasairaala Central Hospital, Satakunta Hospital District, Pori, Finland

- 83 pacientes; control sano x10
- Hospital universitario Turku, Finlandia

TABLE 1 Demographics of the CVID-patients.

CVID patients	
Patients included (N/%)	83 (72.8%)
Mean age (year)	53.7
Male (N)	32
Female (N)	51
Alive (N/%)	66 (79.5%)
Deceased (N/%)	17 (20.5%)
Mean duration of disease (year)	14.8
Immunoglobulin therapy (N/%)	77 (92.8%)
Mean duration of immunoglobulin therapy (year)	13.1
p-IgA mean (g/L)	0.71
p-IgM mean (g/L)	1.15
p-IgG mean (g/L)	4.14
p-IgG4 mean (g/L)	0.13
Vaccine response tested (N/%)	38 (45.8%)
Inadequate response for vaccines (N/%)	31 (81.6%)

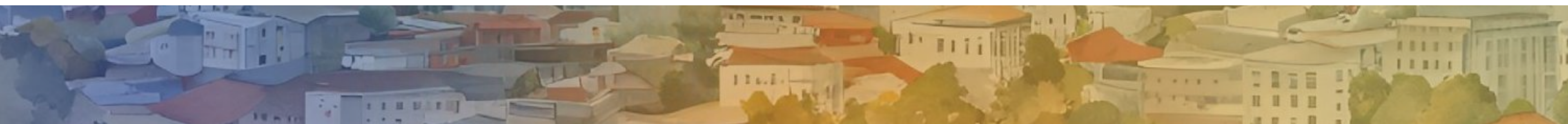


TABLE 2 Combined risk for atherosclerotic disease for CVID patients compared to control group.

Disease	ICD-codes	CVID N (%)	Control N (%)	OR (95% CI)	<i>p</i>
Coronary heart disease	I20–I25	11 (13.25)	50 (6.05)	2.4 (1.2–4.8)	0.015
Peripheral atherosclerosis	I70, I71, I74	8 (9.64)	7 (0.85)	12.5 (4.4–35.4)	<0.001
Cerebrovascular disease	I63–I65, I67, G45, G46	6 (7.23)	31 (3.75)	2.0 (0.8–4.9)	0.133
Any of the above	Any of the above	18 (21.69)	79 (9.55)	2.6 (1.5–4.6)	0.001

Risk factor	CVID		Control		<i>p</i>
	N	N	N	N	
Male gender (N/%)	32 (38.6)	83	319 (38.5)	828	1
Age (year)	53.7 ± 29.5	83	53.6 ± (20.2)	828	0.986 ^a
Hypertension (N/%)	19 (22.9)	83	111 (13.4)	827	0.031 ^b
Diabetes (N/%)	9 (10.8)	83	61 (7.4)	827	0.277 ^b
Any autoimmune disease (N/%)	40 (48.19)	83	119 (14.39)	827	<0.001 ^b
Smoking (N/%)	11 (19.3%)	57	144 (35.0%)	412	0.023 ^b
GHbA1c%	5.60 (5.31–6.21)	30	5.78 (5.5–6.6)	103	0.061 ^c
Cholesterol (mmol/L)	4.61 ± 0.98	43	4.91 ± 1.03	167	0.092 ^a
HDL (mmol/L)	1.39 ± 0.55	41	1.49 ± 0.50	164	0.228 ^a
LDL (mmol/L)	2.50 ± 0.72	41	2.69 ± 0.90	153	0.21 ^a
Triglycerides (mmol/L)	1.35 (1.0–1.8)	44	1.35 (0.95–2.05)	163	0.45 ^c
CRP (mg/L)	6.0 (3.0–18.0)	81	10.5 (3.0–41.0)	428	0.017 ^c

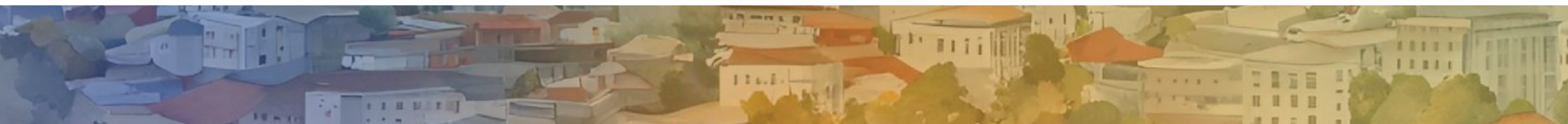
^at-test.

^bFisher's exact test.

^cMann-Whitney test.

TABLE 4 A logistic regression model adjusted for age, sex, smoking status, hypertension, history of any autoimmune disease and median CRP. CVID remained an independent risk factor for atherosclerotic CVDs.

Risk factor	OR (95% CI) N = 469	<i>p</i>
CVID	2.00 (0.90–4.47)	0.002
Age	1.08 (1.05–1.11)	<0.001
Sex	2.82 (1.49–5.37)	0.002
Smoking	1.22 (0.61–2.42)	0.57
Hypertension	2.44 (1.31–4.57)	0.005
Autoimmune diseases	1.31 (0.67–2.54)	0.428
Median CRP	1.0 (0.99–1.01)	0.803





SALUD MENTAL

SALUD MENTAL Y EII

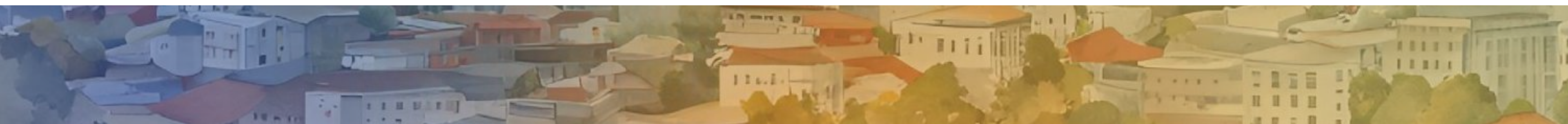
Anxiety in Polish adult patients with inborn errors of immunity: a cross-sectional study

Kinga Grochowalska^{1†}, Marcin Ziętkiewicz^{2†},
Katarzyna Nowicka-Sauer², Mariusz Topolski³,
Ewa Więsik-Szewczyk⁴, Aleksandra Matyja-Bednarczyk⁵,
Katarzyna Napiórkowska-Baran⁶ and Zbigniew Zdrojewski¹

Results: Thirty-eight (36.2%) patients had anxiety symptoms (HADS-A ≥ 8); 14 (13.3%) patients had severe anxiety (score ≥ 11), and 24 (22.9%) had moderate anxiety (score of 8–10). Patients with poor sleep quality, higher pain frequency, younger age, and no fixed income had higher anxiety scores than others. Emotional and cognitive representations of illness were positively correlated with anxiety levels. Intense anxiety was related to more negative illness perception, higher helplessness, lower illness acceptance, and lower perceived benefits.

Variable		
Anxiety (HADS)		
No anxiety symptoms (< 8 points)		6
Moderate anxiety symptoms (8–10 points)		2
Severe anxiety symptoms (≥ 11 points)		1
Sleep quality (PSQI)		
Good (< 5 points)		5
Poor (≥ 5 points)		4

IEI, inborn errors of immunity; Ig, immunoglobulin; IVIg, intravenous SCIg, subcutaneous immunoglobulin; HADS, hospital anxiety and depression; PSQI, Pittsburgh sleep quality index; n, number.



SALUD MENTAL Y EII

Perceived Sleep Quality in Individuals with Inborn Errors of Immunity

Kerri L. Sowers¹ · Adel Sawaged¹ · Brian Bowen¹

Received: 4 January 2023 / Accepted: 17 March 2023
 © The Author(s), under exclusive licence to Springer Science+Business Media, LLC, part of Springer Nature 2023

Table 3 Sleep quality demographics

Sleep diagnosis % (n)	n = 548
None	51.8 (284)
Sleep apnea	28.5 (156)
Insomnia	20.6 (113)
Restless leg syndrome	12.4 (68)
Other ^a	13.3 (73)
Symptoms of poor/disrupted sleep % (n)	n = 548
Snoring	43.2 (237)
Bruxism	41.4 (227)
Vivid dreams	35.0 (192)
None	17.7 (97)
Nightmares	17.0 (93)
Sleep talking	16.2 (89)
Other ^b	8.2 (45)

Table 2 Health demographics

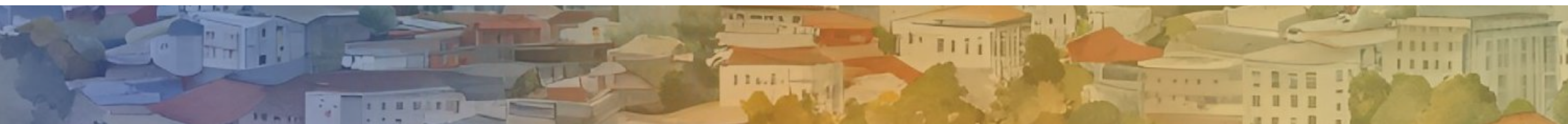
Participants	n = 548
Type of IEI % (n)	
CVID	83.8 (459)
SCID	0.5 (3)
SAD	3.6 (20)
IgG subclass deficiency	5.7 (31)
XLA	1.3 (7)
Selective IgA deficiency	0.7 (4)
Complement deficiency	0.2 (1)
CGD	0.4 (2)
Other	2.6 (14)
Unsure	1.3 (7)
Time diagnosed % (n)	
< 1 year	7.8 (43)
1–5 years	35.0 (192)
6–10 years	26.1 (143)
11–15 years	16.1 (88)
> 15 years	15.0 (82)
Treatment % (n)	
No treatment	6.9 (38)
IVIG	33.4 (183)
SCIG	58.4 (320)
Prophylactic antibiotics	9.3 (51)
Other	2.0 (11)

Puntaje: 43 (dificultad moderada)

Table 4 Quality of life issues relating

Chronic pain interferes with sleep %
Yes
No
Unsure
Mental health % (n)
Anxiety
Depression
Other mental health diagnosis
No mental health diagnosis
Smoking history % (n)
Never smoked
10 years or less
More than 10 years
Work Shifts % (n)
Not working/working non-shift hours
Traditional day shifts
Other ^c

Other^c = Late afternoon/early evening shifts, or very early morning shifts





¡GRACIAS!