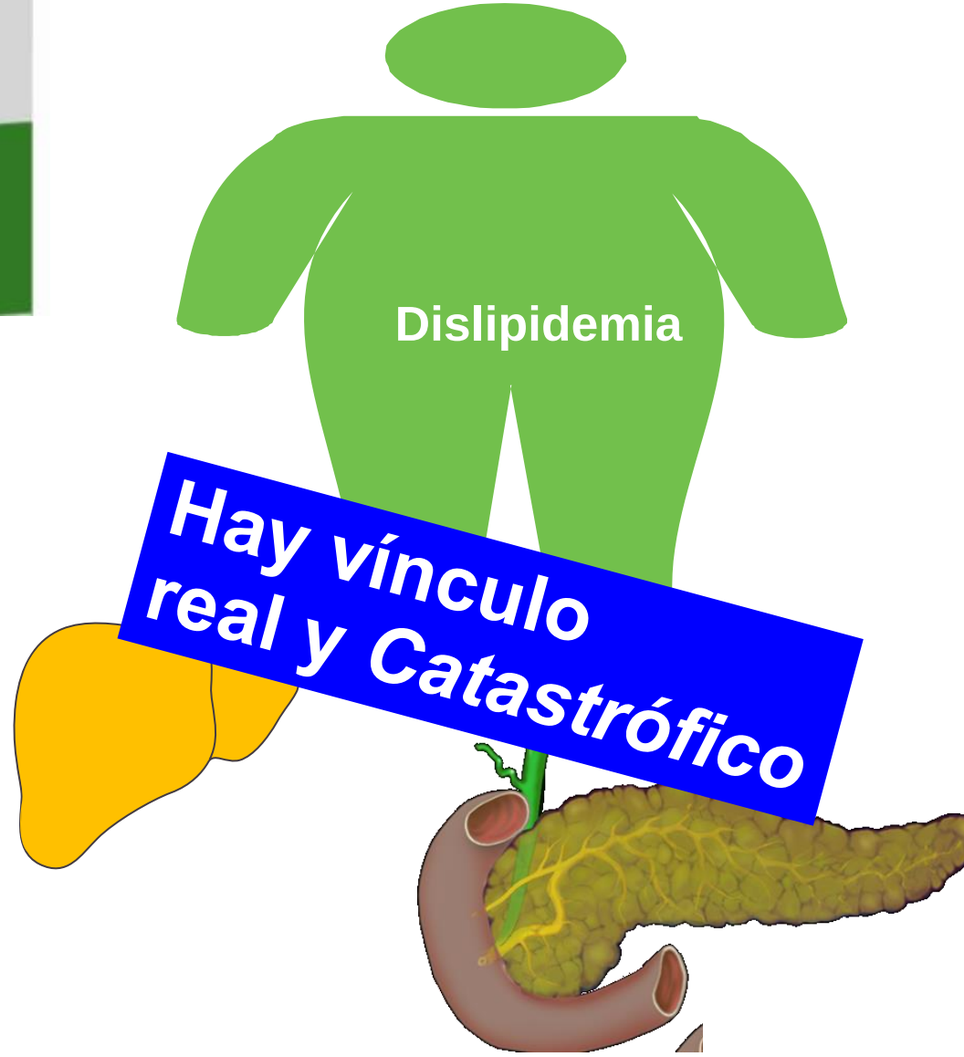
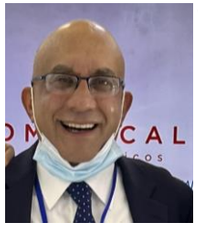


Obesidad y dislipidemias: impacto en hígado y páncreas .



William Otero R, MD, FAGA, FACP
Profesor Titular de Medicina
Universidad Nacional de Colombia
Hospital Universitario Nacional

Canal YouTube “William Otero Gastroenterólogo”

Conflicto de intereses
Conferencista, Bristol
Takeda, Abbott, Tecnoquimica
Tecnofarma, Menarini, Procaps

Esta actividad es asuspiciada por
PTC Therapeutics sin injerencia en su contenido

Obesidad

13%	Mundo
42.4%	USA

“Enfermedad crónica, neuro-comportamental recurrente, incurable, un aumento de la grasa corporal, Disfunción tisular, con efectos adversos metabólicos, bioquímicos, sicosociales”

IMC ≥30 Kg M^2: grasa o músculos?

30-34.9 Kg/M^2, cintura
>102 cm Hombres
> 88 cm mujeres

% Grasa
> 25 Hombres (8-19)
>35 Mujeres (21-35)

Jirapinyo P, Am J Gastroenterol 2021; Epub ahead Abril 6
Obesity Medicine Association 2020

Obesidad

4.5 millones muertes 2013

Lauby-Secretan B, IARC N Engl J Med 2016;375:794-8

Osteoporosis

Artrosis rodilla

Artrosis Cadera

Cáncer de Seno

Pancreatitis Aguda

Pancreatitis Crónica

Cáncer de Utero

Reflujo Gastroesofágico

Cáncer de Hígado

Apnea Del sueño

Cáncer de Páncreas

Insuf Rena

de

Hipe Arter

Trom cere

de o

Infarto Miocardio

de Riñón

Infarto Intestinal

Primera causa de cirrosis
Primera causa de trasplante de hígado en mujeres

Diabetes Mellitus

Resistencia Insulina

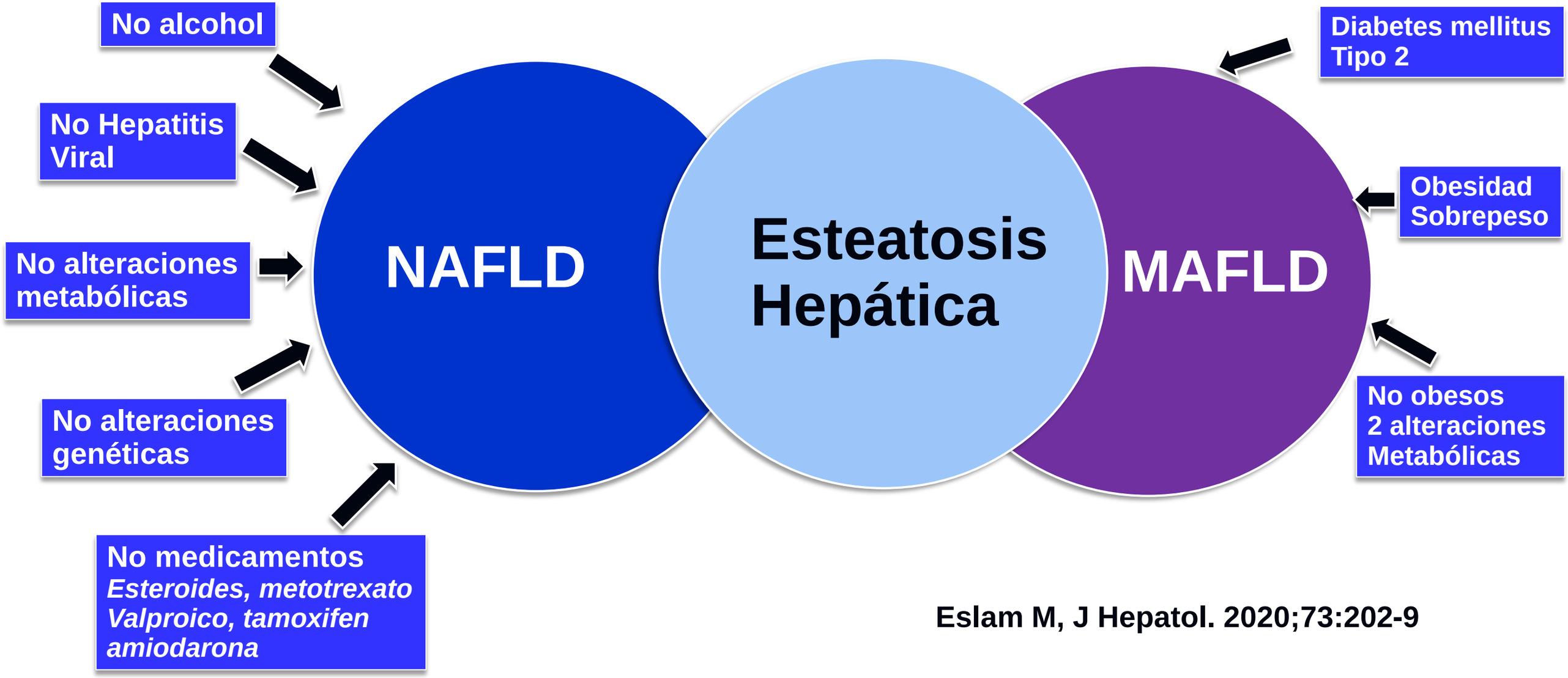
Ovario Poliquístico

Diarrea Crónica

IMC > 30
Vive 9 años menos

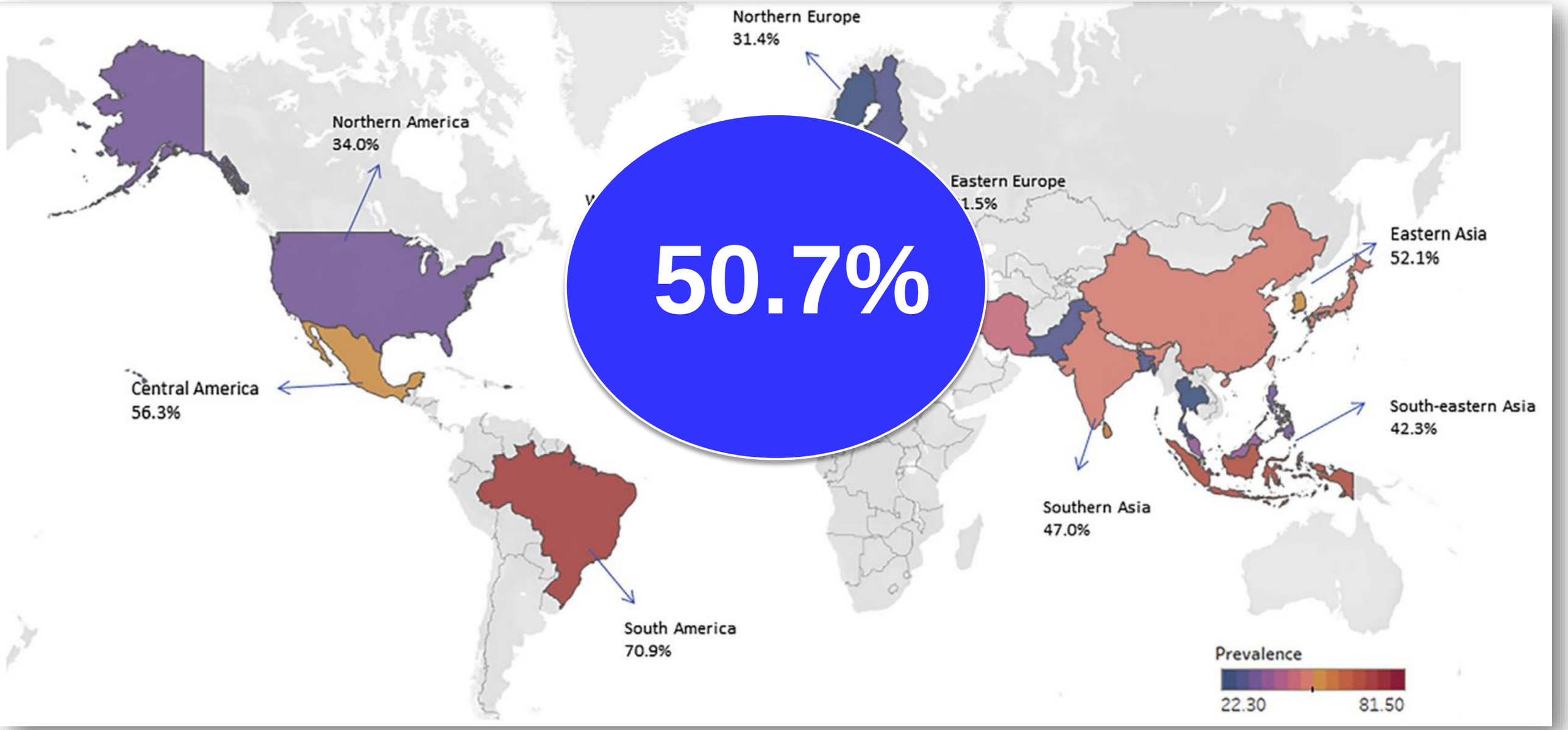
IMC > 40
Vive 13 años menos

Withlock G, Lancet 2006;368:666-78
Brolin RE, JAMA 2003;289:187-93



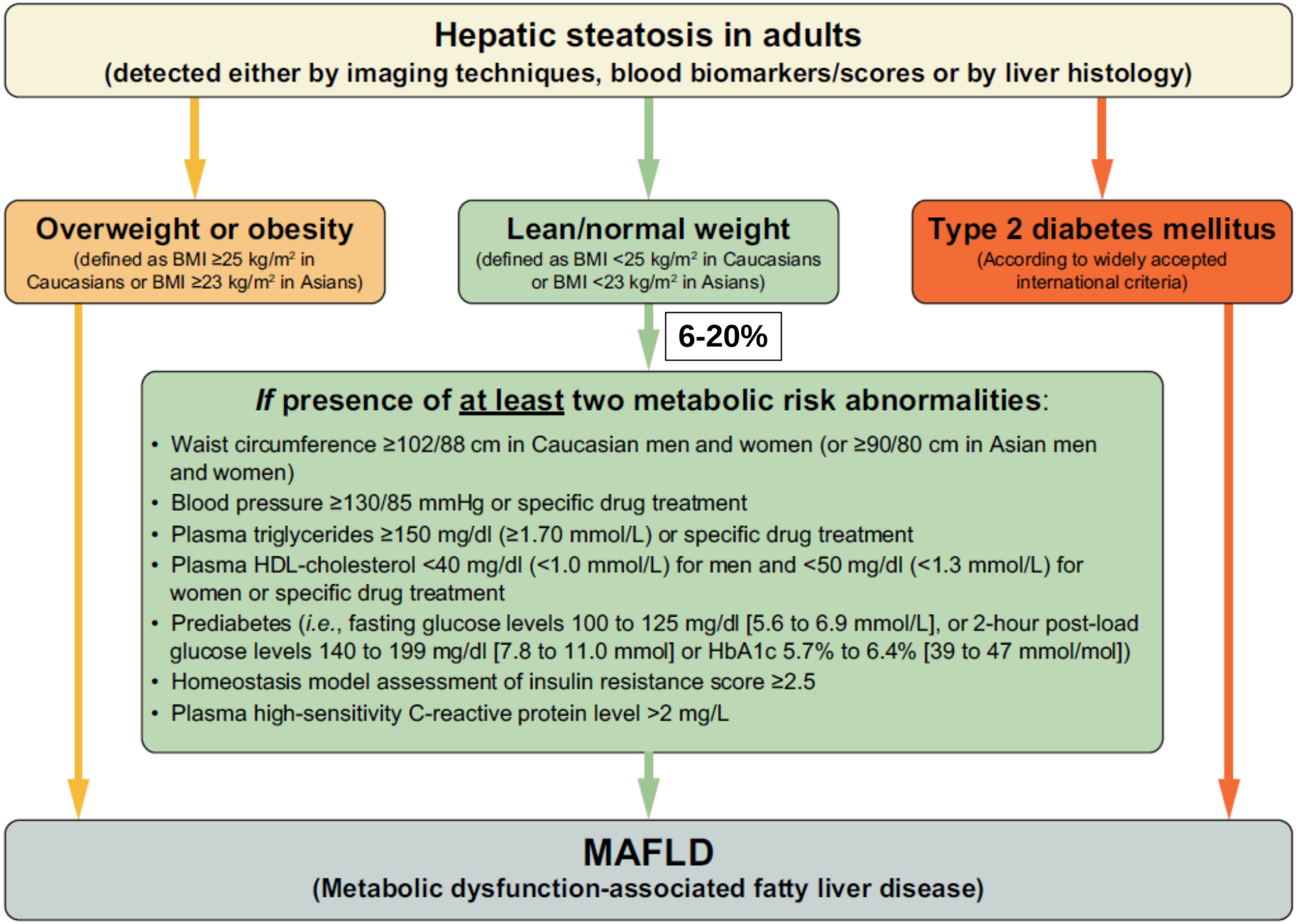
Eslam M, J Hepatol. 2020;73:202-9

Prevalencia global de MAFLD sobrepeso/obesidad



Liu J, et al. Clin Gastroenterol Hepatol 2022;20:573-82

Diagnóstico Positivo



Eslam M, J Hepatol. 2020;73:202-9

Dual aetiology fatty liver disease (concomitant MAFLD and other liver disease).

Meeting the criteria for a diagnosis of MAFLD

Plus

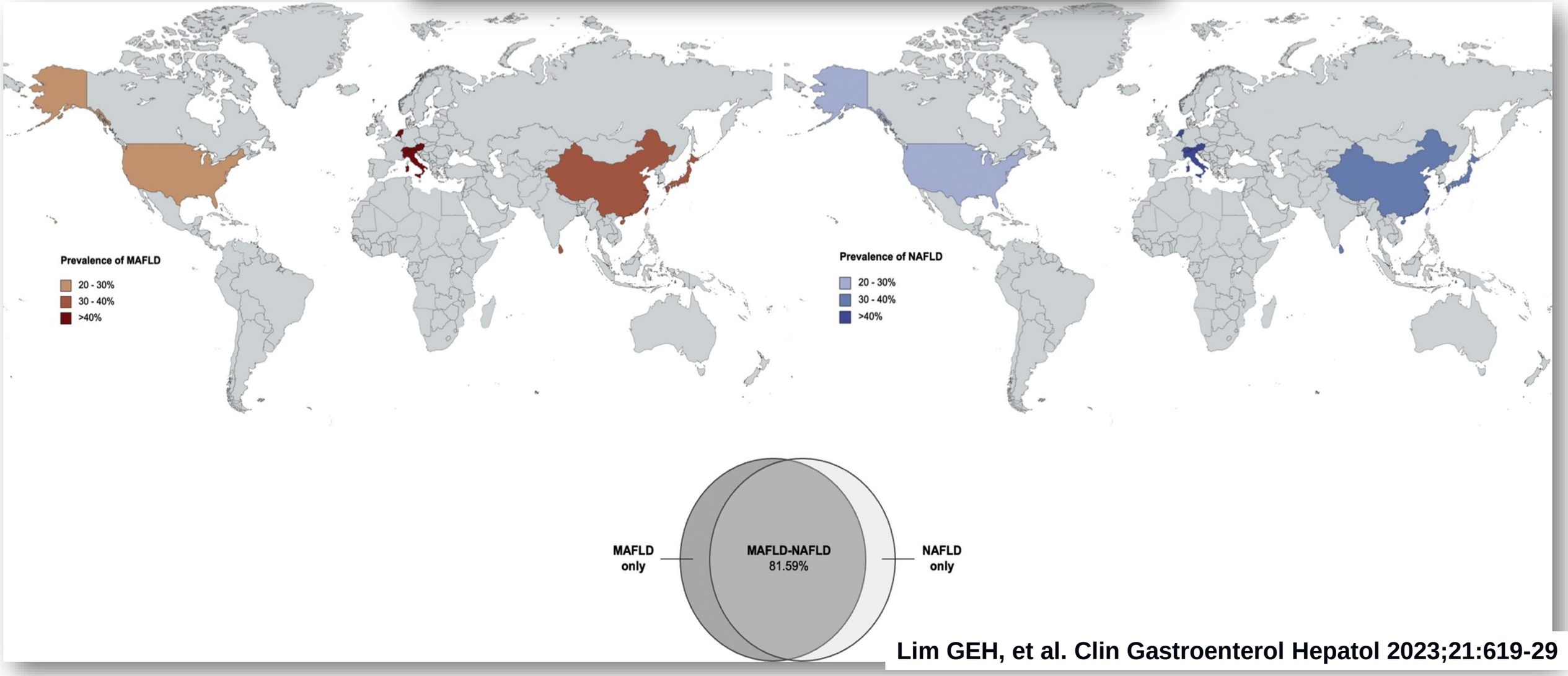
Any other cause for liver disease *e.g.*, alcohol-use disorder defined as consumption of >3 drinks per day in men and >2 drinks per day in women, or binge drinking (defined as >5 drinks in males and >4 drinks in females, consumed over a 2 hour period)*, as defined by the National Institute of Alcoholism and Alcohol Abuse[47,62], viral infection (HIV, HBV and HCV), autoimmune hepatitis, inherited liver disorders, drug-induced liver injury or other known liver disease

Puede empeorar otras enfermedades Hepáticas crónicas Pre existentes, Autoinmune, hepatitis virales, etc

Eslam M, J Hepatol. 2020;73:202-9

An Observational Data Meta-analysis on the Differences in Prevalence and Risk Factors Between MAFLD vs NAFLD

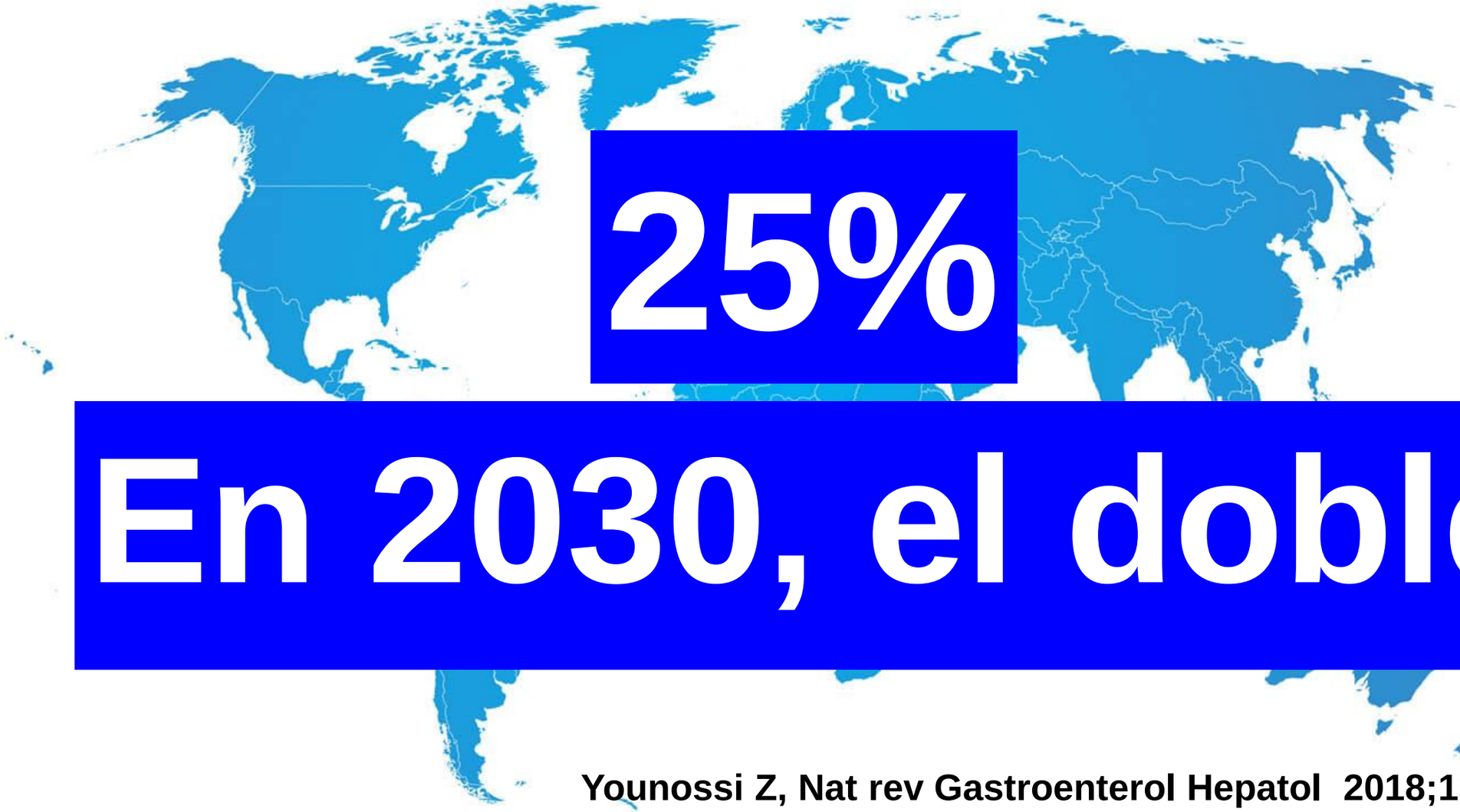
Grace En Hui Lim,[*,a] Ansel Tang,[‡,a] Cheng Han Ng,[‡,a] Yip Han Chin,[‡] Wen Hui Lim,[‡] Darren Jun Hao Tan,[‡] Jie Ning Yong,[‡] Jieling Xiao,[‡] Chloe Wen-Min Lee,[‡] Mark Chan,[‡,§] Nicholas WS. Chew,[§] Eunice Xiang Xuan Tan,[‡,||,¶] Mohammad Shadab Siddiqui,[#] Daniel Huang,[‡,||,¶] Mazen Noureddin,[**] Arun J. Sanyal,[#,b] and Mark D. Muthiah[‡,||,¶,b]



Lim GEH, et al. Clin Gastroenterol Hepatol 2023;21:619-29

Singh SP, J Clin Exper Hepatol 2023;13:518-523

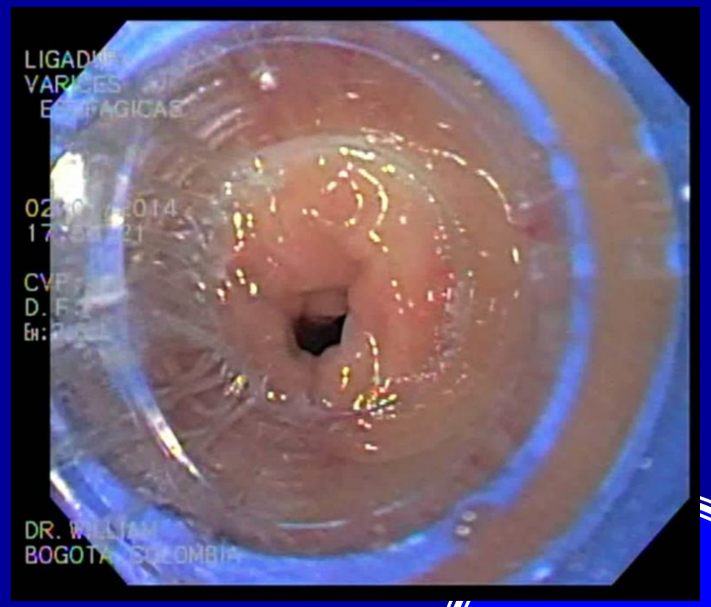
NAFLD, prevalencia



25%

En 2030, el doble

Younossi Z, Nat rev Gastroenterol Hepatol 2018;15:11-20



Cirrosis Criptogènica

ALT

MAFLD

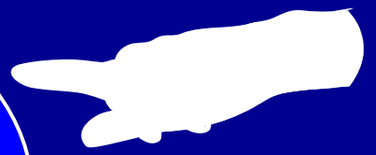
Principal Causa Hepatitis Crónica

Trasplante hepático
1ª Causa Mujeres
2ª Global

Samji NS, J Clin Exp Hepatol 2019;9:497-505

NAFLD/NASH

Enfermedad Cardiovascular 13-30%



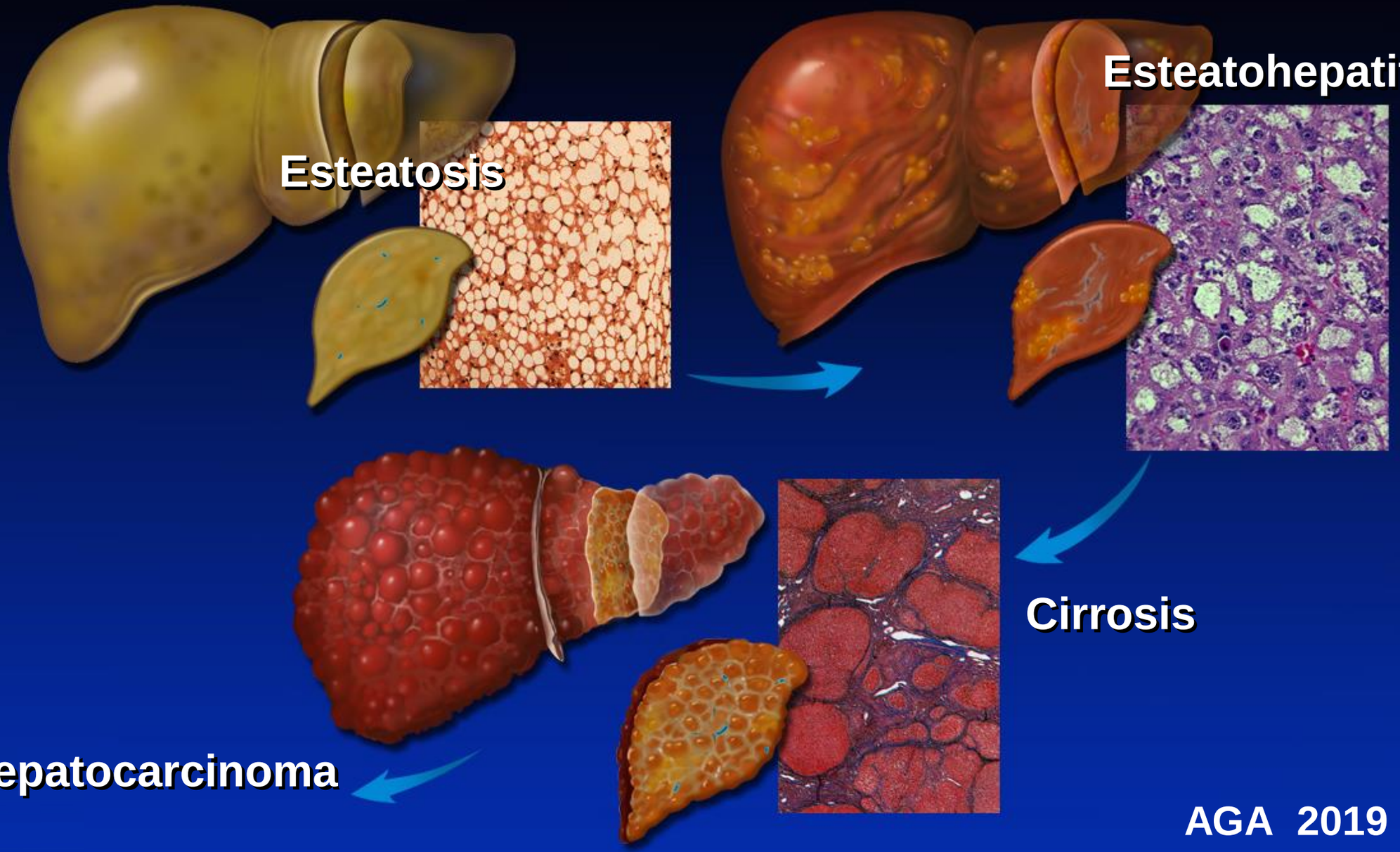
Principales Causas De muerte

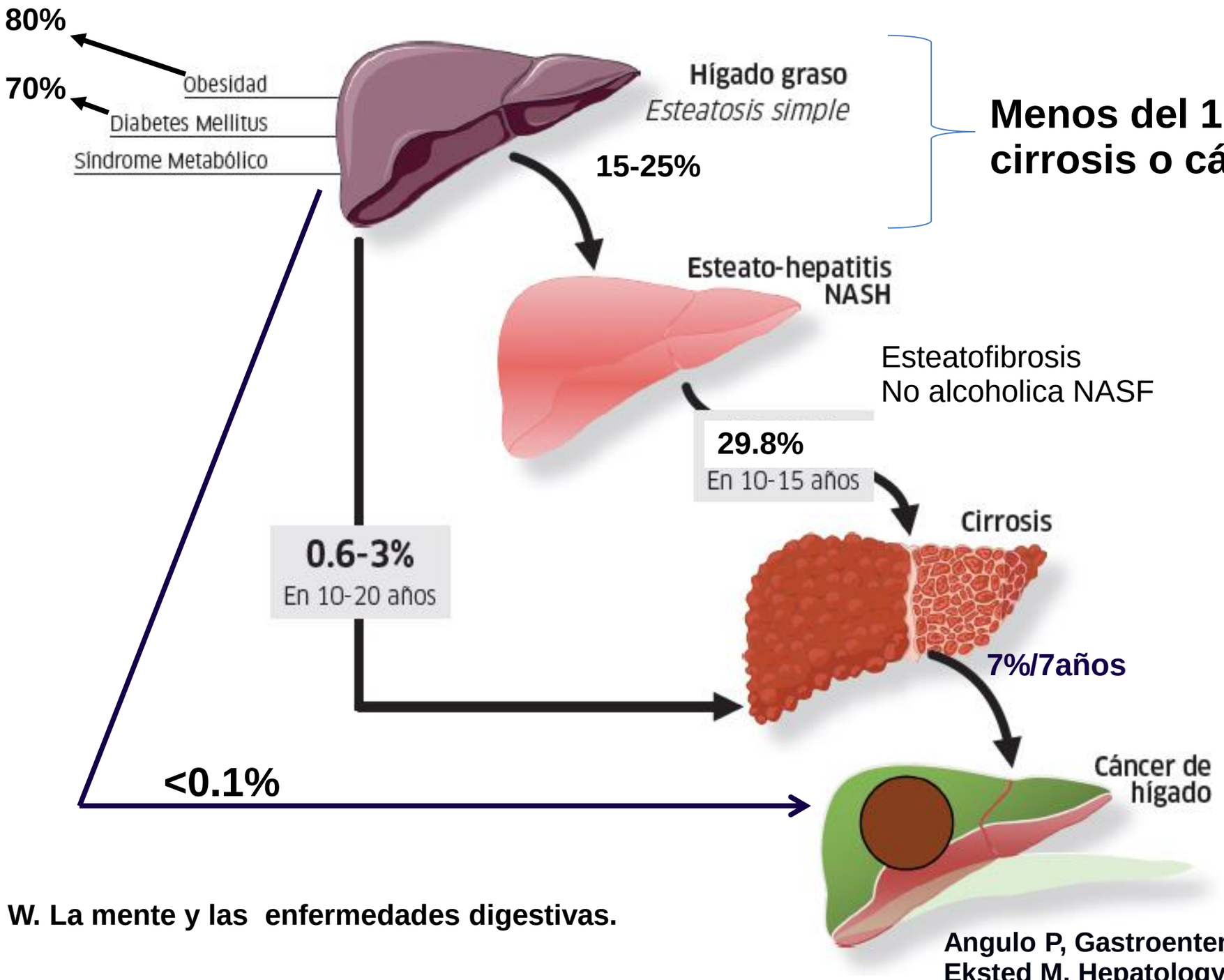
Tumores 6-28%

Hígado 2.8-19%

Pagada MR, Clin Liv Dis 2012;16:487-504

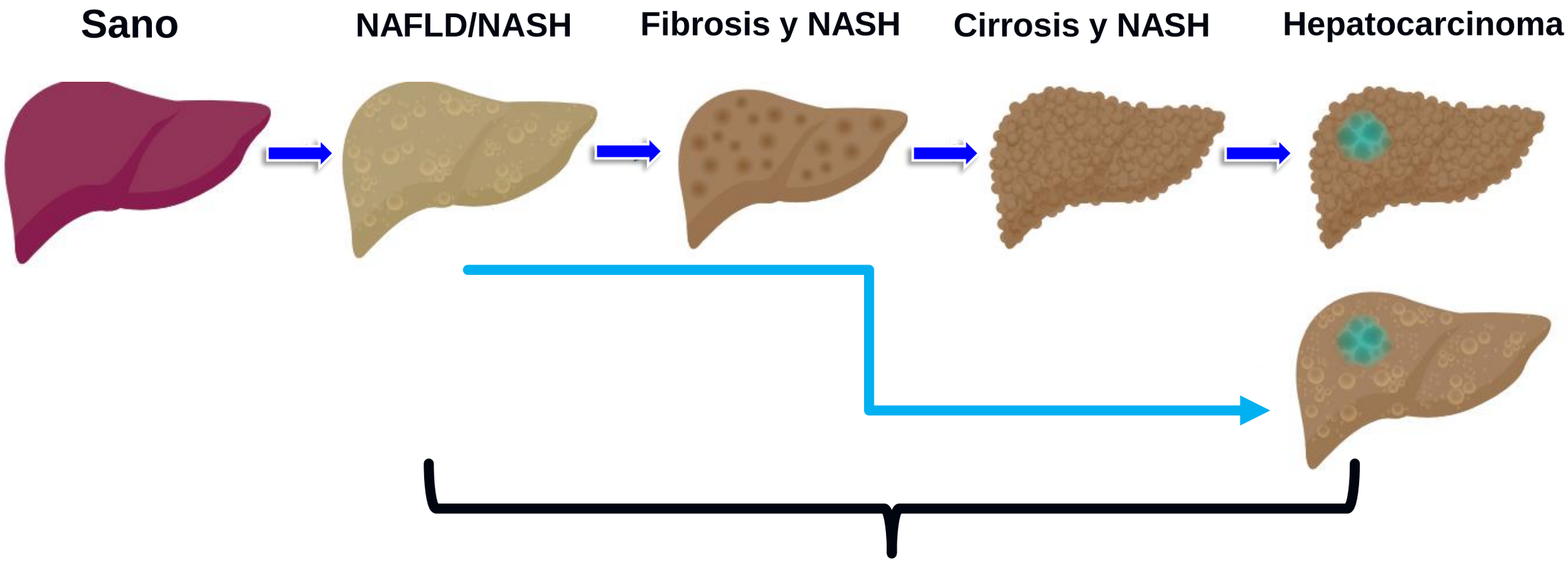
NAFLD/MAFLD
Espectro histológico





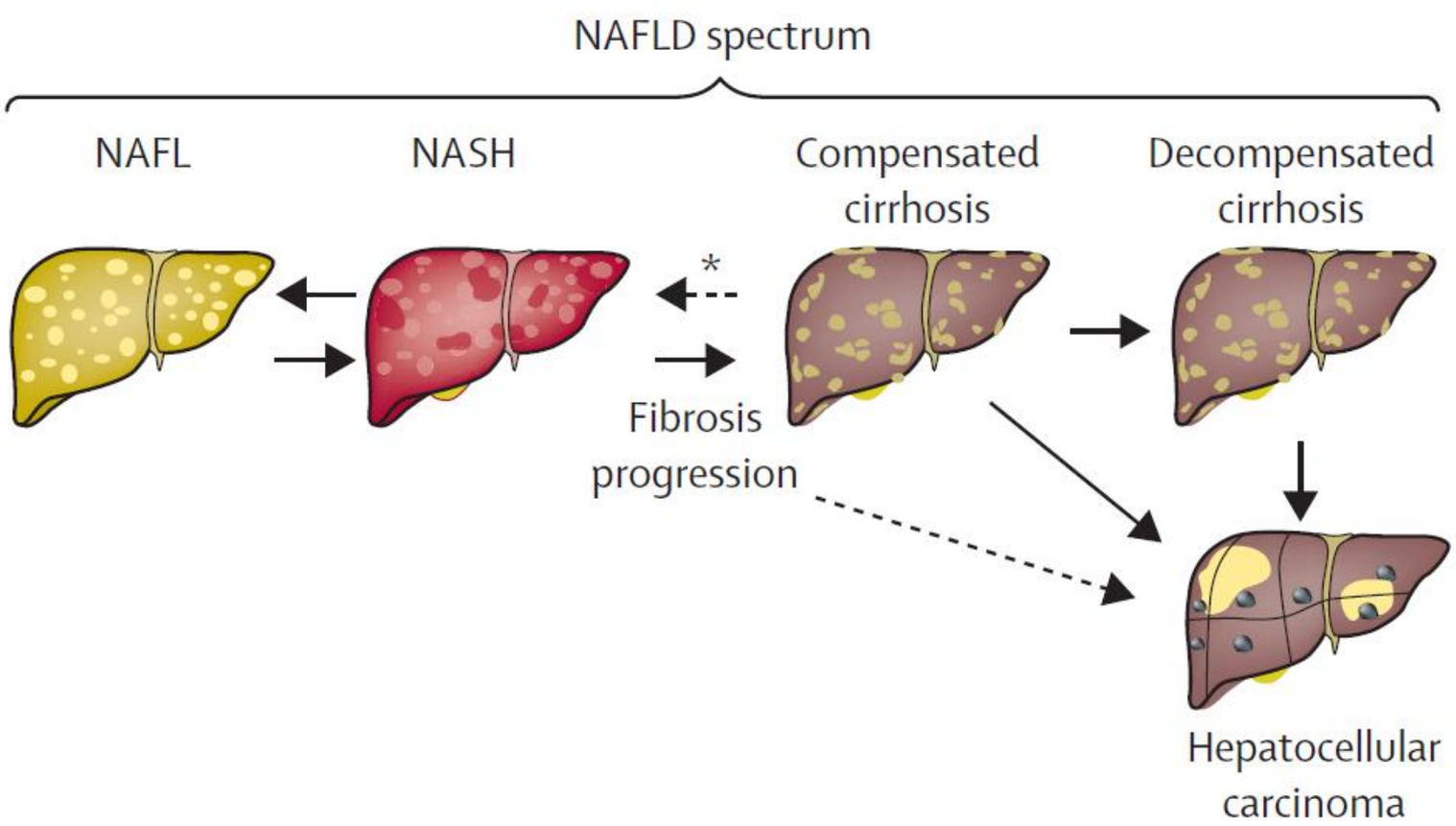
Alvarez P, Otero W. La mente y las enfermedades digestivas. Bogotá 2016

Angulo P, Gastroenterology 2015;149:389-97
Eksted M, Hepatology 2015;61:1547-54



Riesgo aumentado de diabetes tipo II
Riego aumentado de enfermedad cardiovascular

Tsochatzis EA, Nature 2022;19:151-2



Factors associated with NAFLD and NASH progression

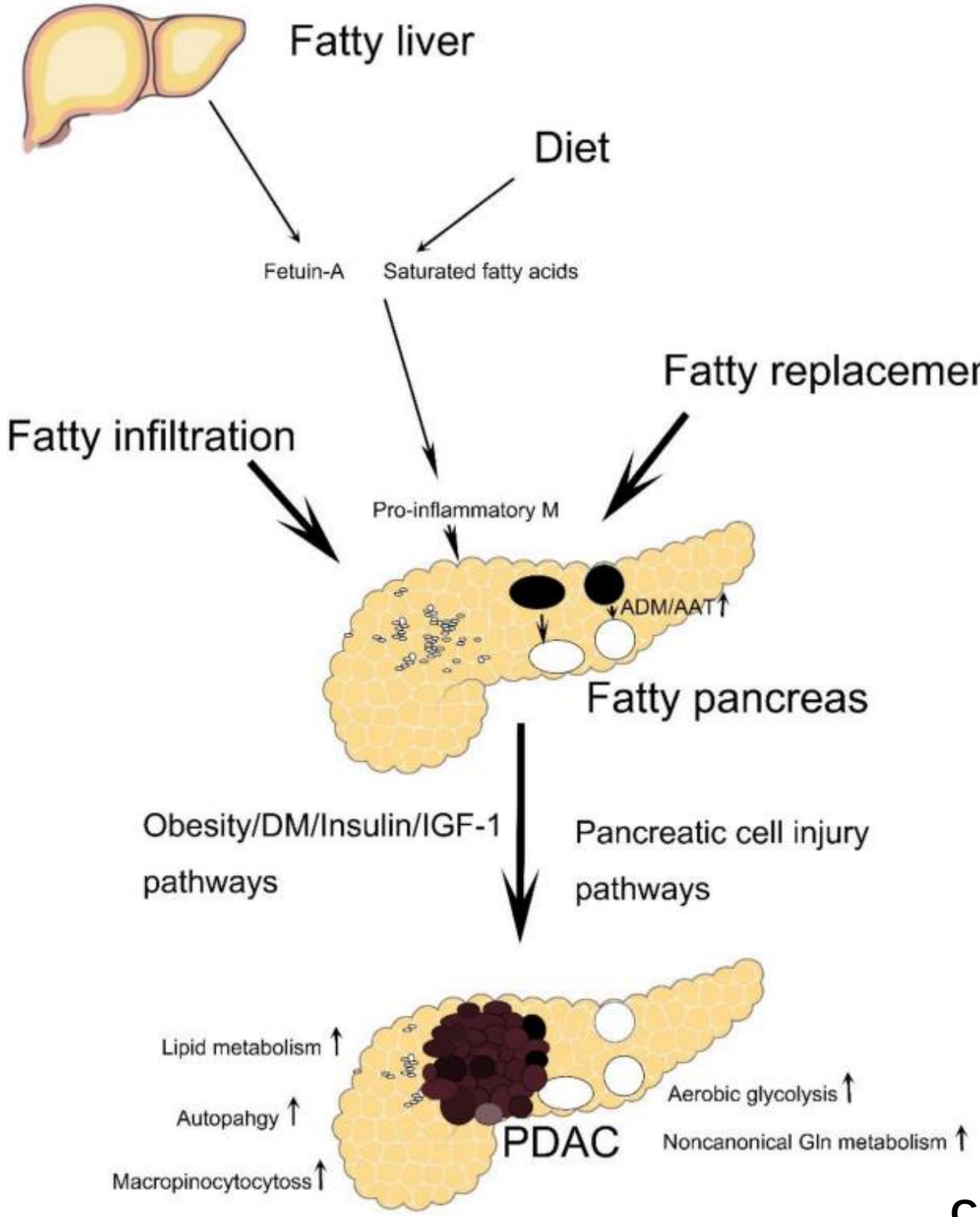
Comorbid illness
- Type 2 diabetes
- Insulin resistance
- Dyslipidaemia
- Obesity
- Hypertension
- Hypopituitarism

Genetic factors
- *PNPLA3*
- *TM6SF2*
- *GCKR*
- *MBOAT7*
- ***HSD17B13***

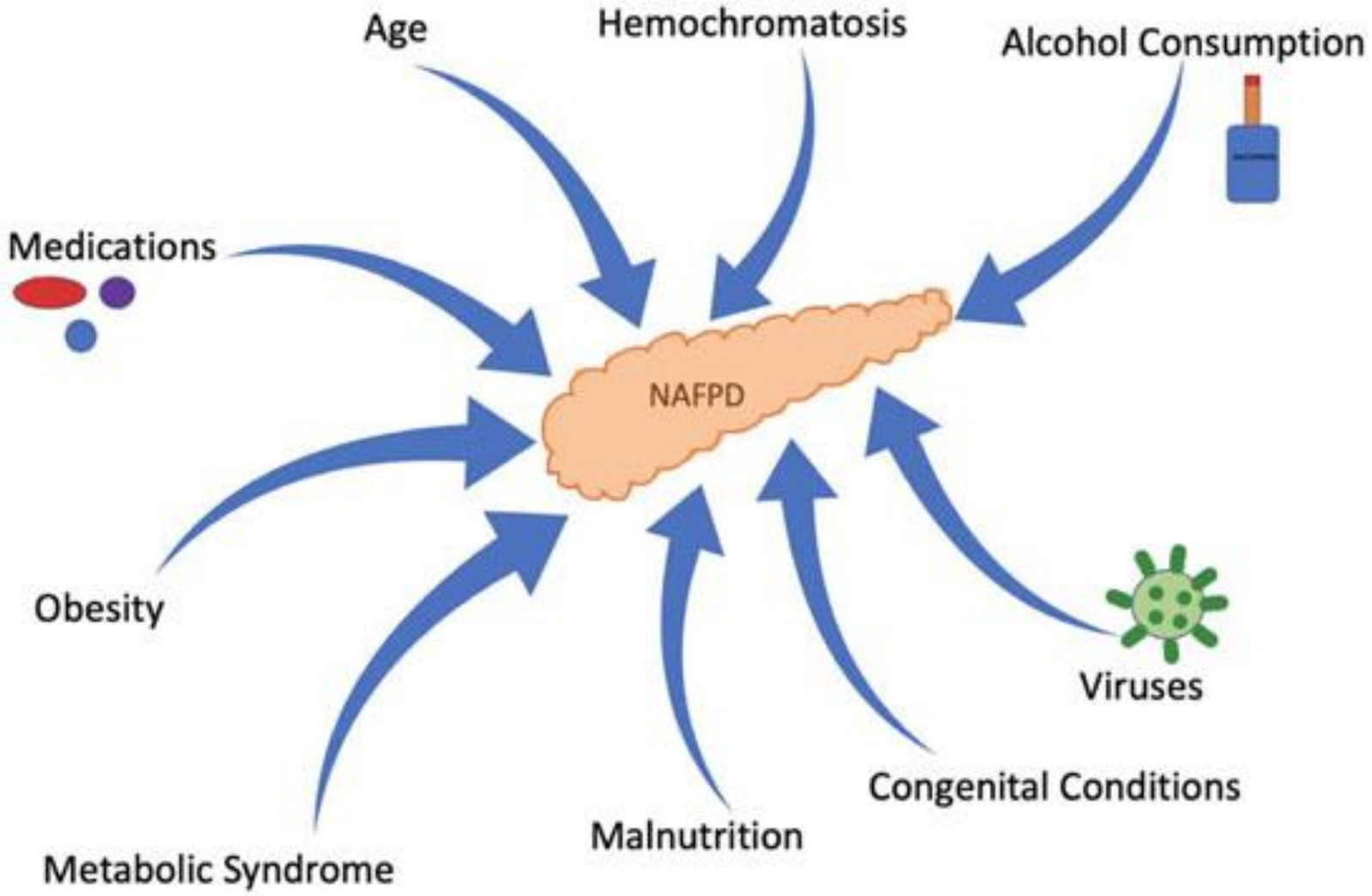
Environmental factors
- Fructose
- Cholesterol
- Alcohol
- **Exercise**
- **Coffee**

Epigenéticos
Microbioma

Powell EE, Lancet. 2021 Jun 5;397(10290):2212-24.

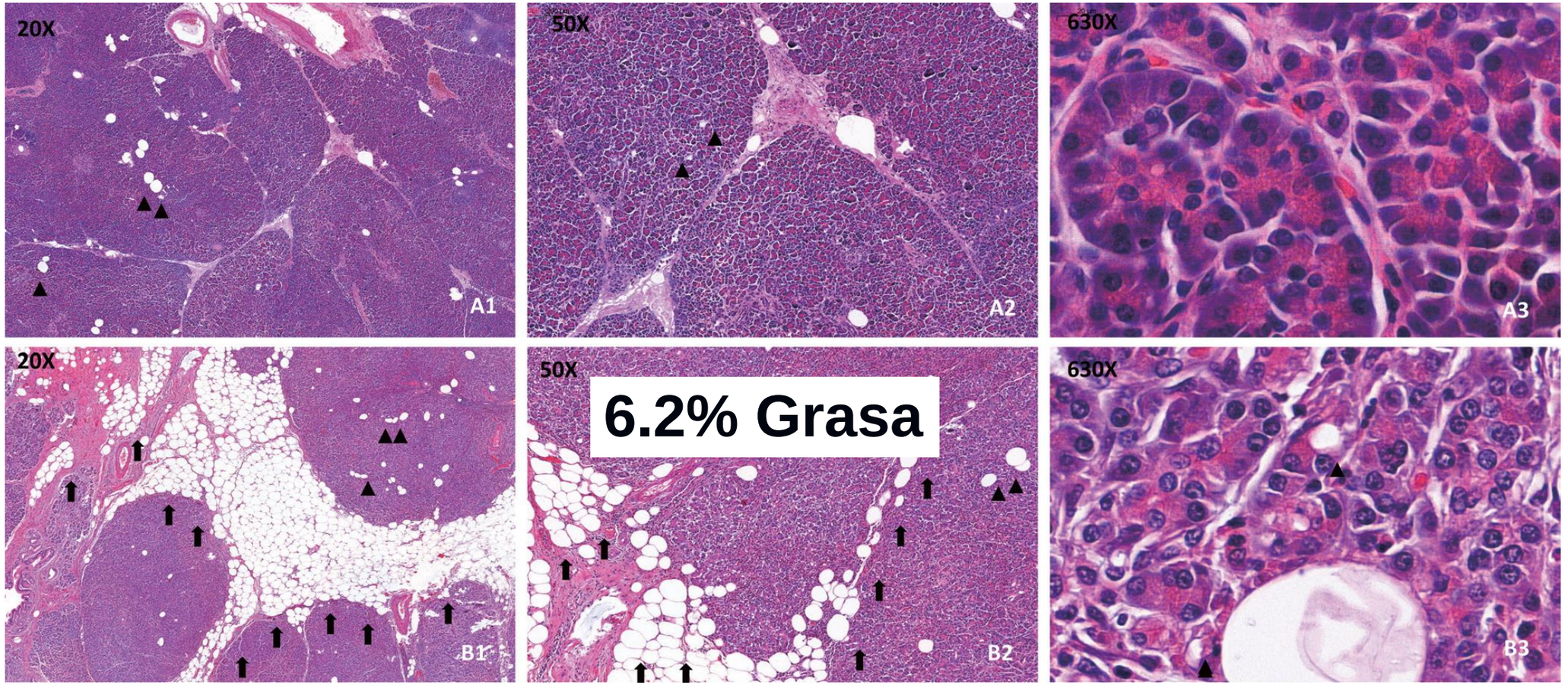


Chang MJ, Biomedicines 2022;10:692.



Pathogenesis of NAFPD.

Páncreas graso



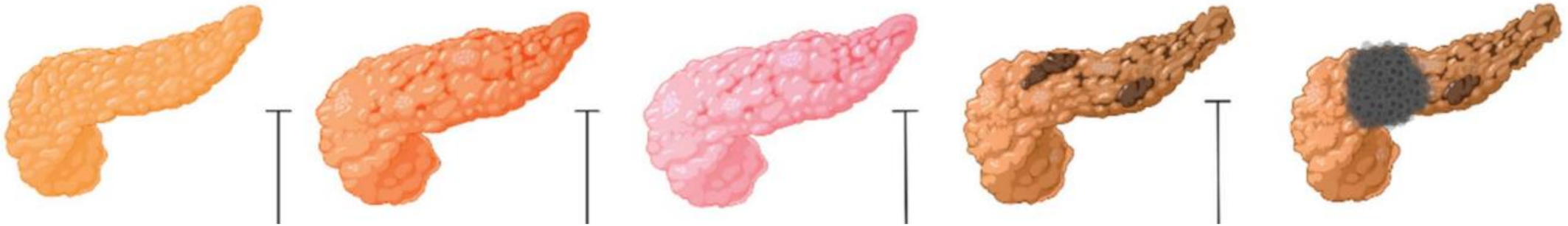
11-35% Asiáticos
61% clínica obesos
28% USA USE

Singh RG, Metabolism 2017;69:1–13
Rugivarodom M, J Clin Trans Hepatol 2022;10:1229-39

Depósito Graso en páncreas

Reversible | Reversible | Irreversible | Irreversible

Páncreas normal | Pancreatitis aguda | Páncreas graso | Pancreatitis crónica | Cáncer de páncreas



Pancreatitis aguda
Predisposición

Cálculos biliares
Alcohol
Iatrogénica
Otros

Transdiferenciación
Acinar-Adiposito

Meta inflamación

Cambios genéticos
En identidad celular

MAFLD | **Cirrosis** | **Hepato carcinoma**

Primer Hit | **Segundo Hit** | **Carcinogénesis**

Schepis T, Am J Gastroenterol 2023;118:910–911

Intrapancreatic, Liver, and Skeletal Muscle Fat Depositions in First Attack of Acute Pancreatitis Versus Health

Juyeon Ko, MSc, PhD[1], Zena Al-Ani, MBChB, BMedSc(Hons)[1], Kieran Long, MBChB[1], Conor Tarrant, MBChB[1], Loren Skudder-Hill, MBBS, BMedSc(Hons)[1] and Maxim S. Petrov, MD, MPH, PhD[1]

Ko J, Am J Gastroenterol 2022;117:1693–1701

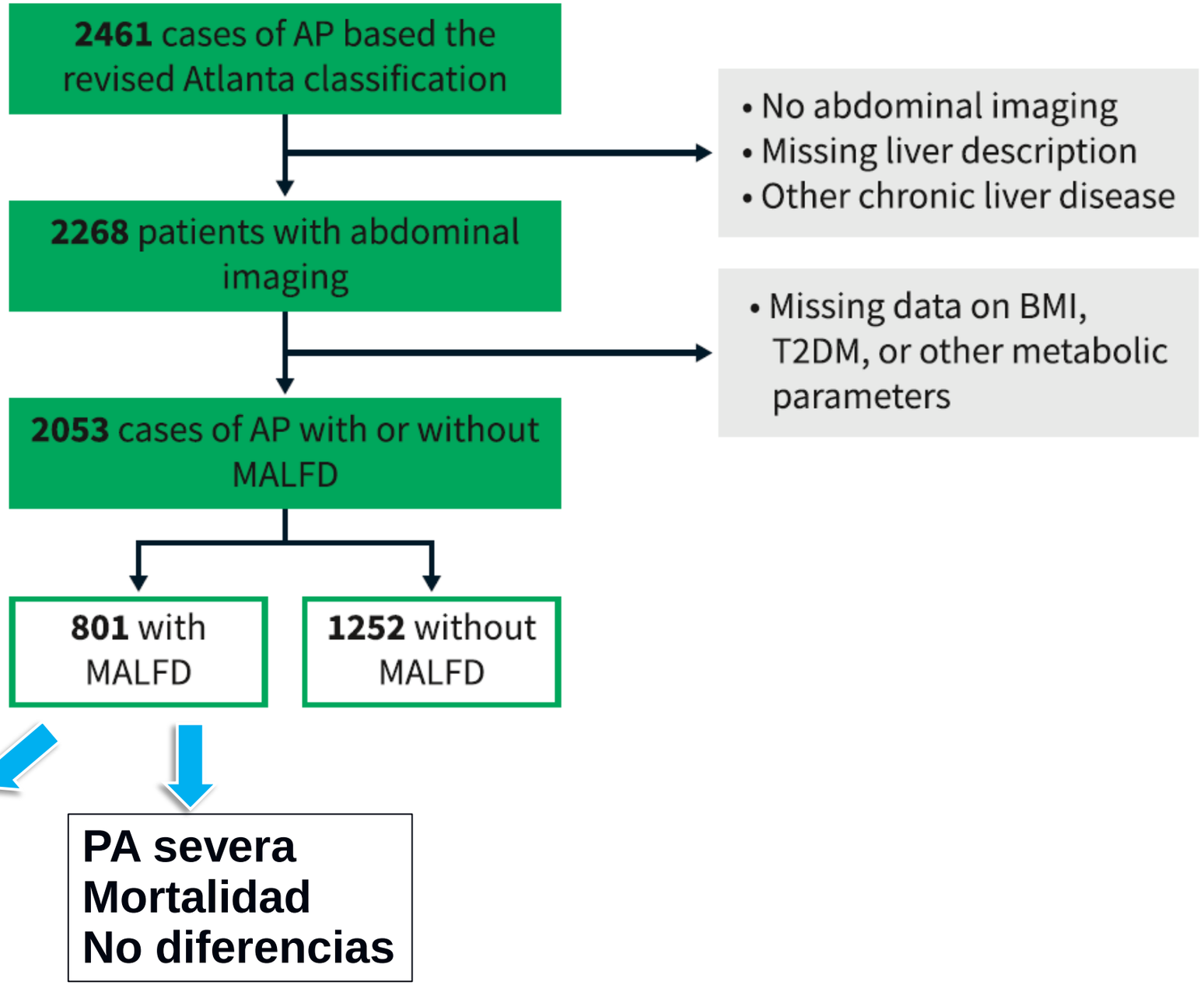
ORIGINAL ARTICLE

ueg journal WILEY

Metabolic-associated fatty liver disease is associated with acute pancreatitis with more severe course: Post hoc analysis of a prospectively collected international registry

Szilárd Váncsa[1,2,3] | Zoltán Sipos[1,4] | Alex Váradi[1,5,6] | Rita Nagy[1,2,7] | Klementina Ocskay[1,7] | Félix Márk Juhász[1,7] | Katalin Márta[2,3] | Brigitta Teutsch[1,2] | Alexandra Mikó[1,8] | Péter Jenő Hegyi[1,2,3] | Áron Vincze[9] | Ferenc Izbéki[10] | László Czakó[11] | Mária Papp[12] | József Hamvas[13] | Márta Varga[14] | Imola Török[15] | Artautas Mickevicius[16,17] | Bálint Erőss[1,2,3] | Andrea Párniczky[1,2,7] | Andrea Szentesi[1,18] | Gabriella Pár[1,9] | Péter Hegyi[1,2,3,18] | on behalf of the Hungarian Pancreatic Study Group

Váncsa S, et al. United European Gastroenterol J. 2023;11:371–382.



PA Moderamente severa
OR 1.39 (IC95% 1.05-1.84)

PA severa
Mortalidad
No diferencias

Váncsa S, et al. United European Gastroenterol J. 2023;11:371–382.

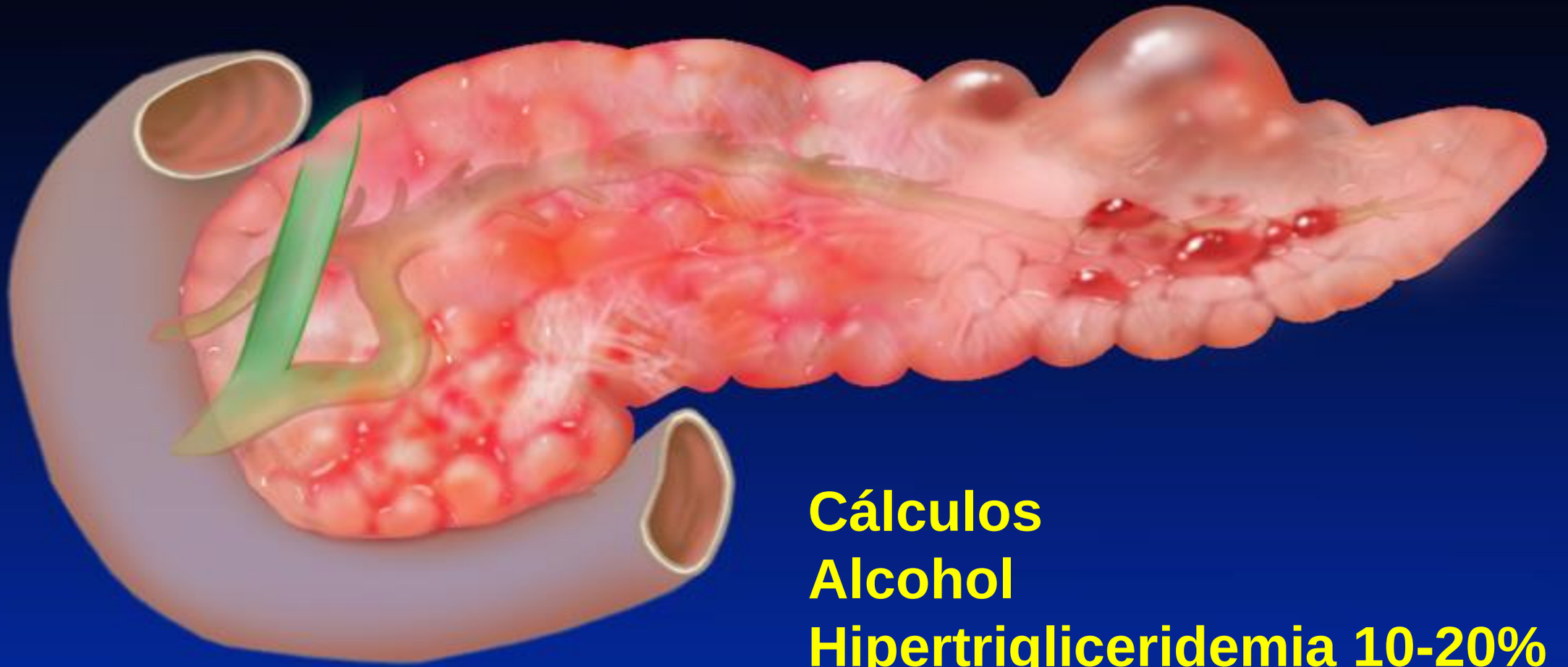
Pancreatitis aguda

Dolor abdominal

Amilasa/Lipasa 3 veces

Imágenes

Banks PA,. Gut 2013;62:102–11.



Cálculos

Alcohol

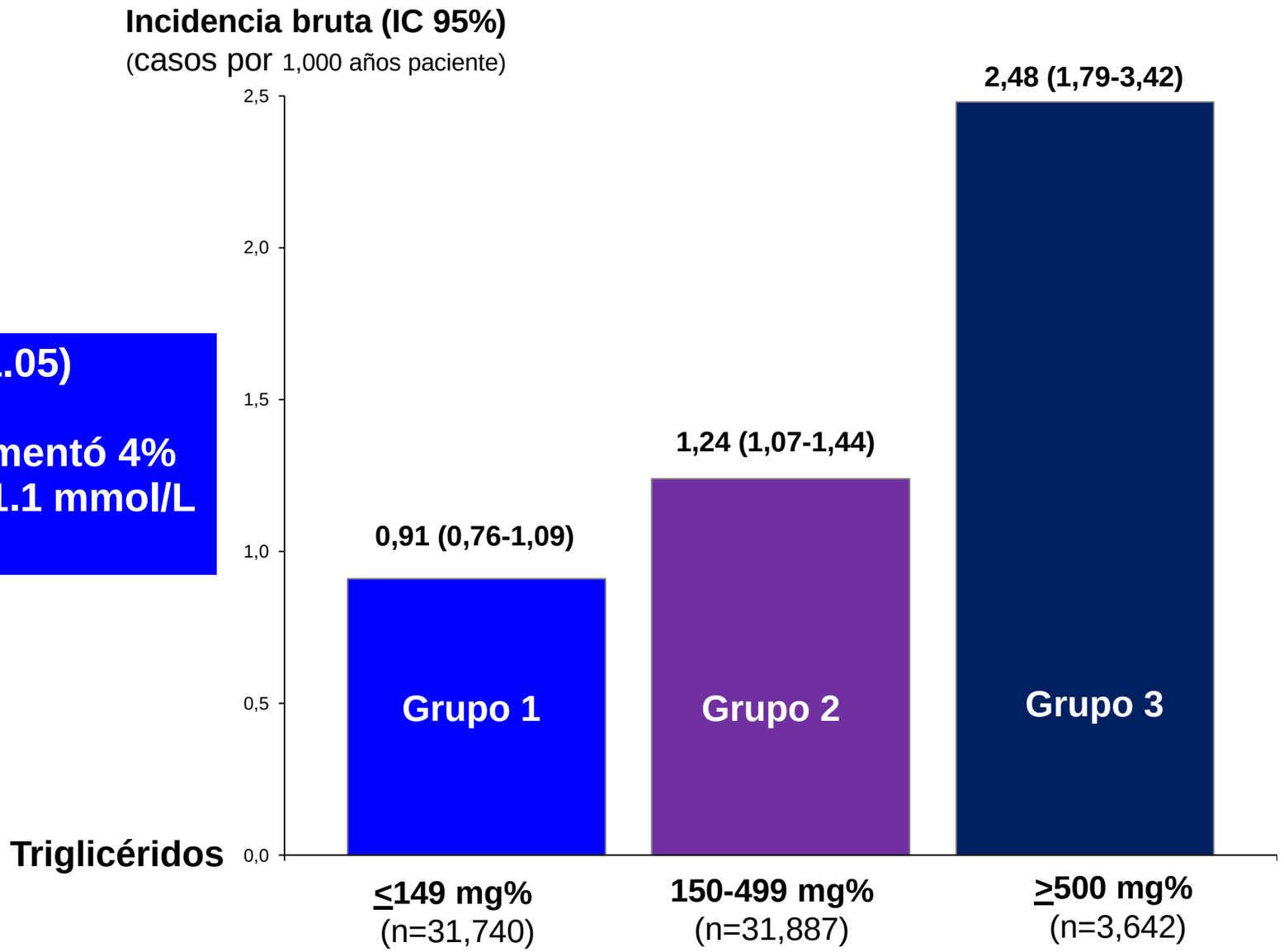
Hipertrigliceridemia 10-20%

Gelrud A, et al. Expert Rev Cardiovasc Ther. 2017;15:879-87.

El riesgo de pancreatitis aumenta con los niveles de triglicéridos

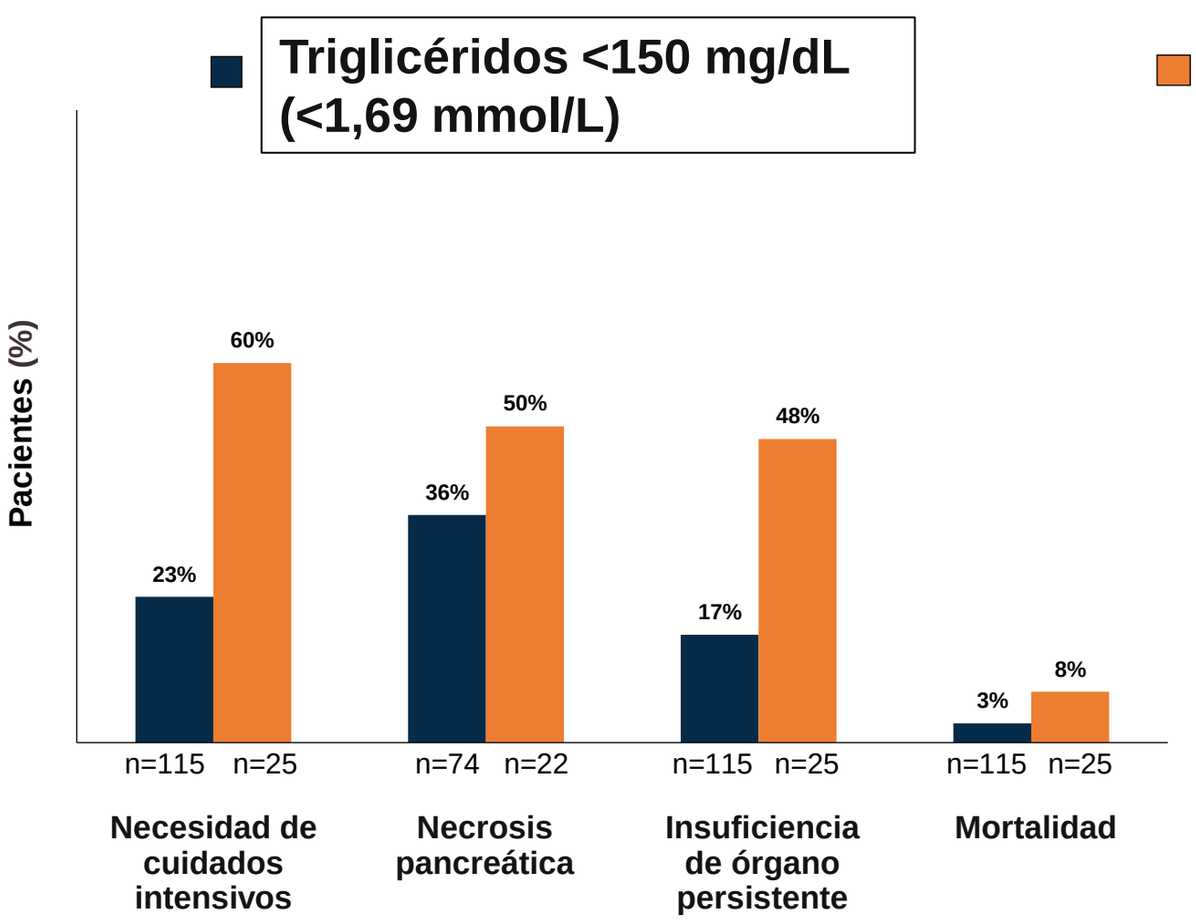
OR 1.04 (IC 95% 1.02-1.05)

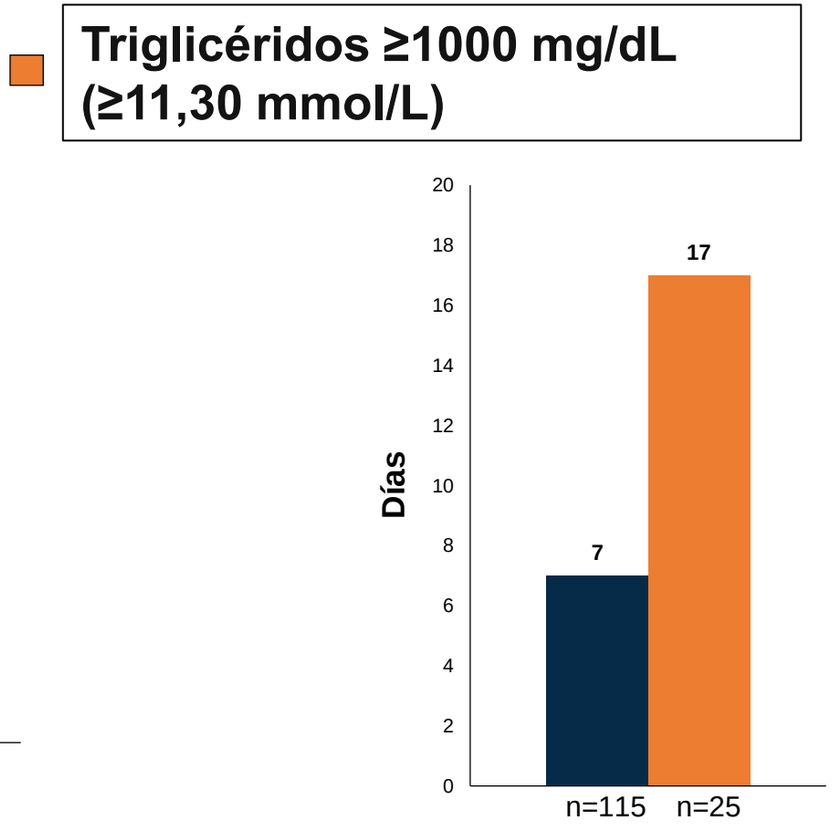
Pancreatitis aguda aumentó 4% por cada aumento de 1.1 mmol/L del valor de TG



Murphy M, et al. JAMA Intern Med. 2013;173:162-164

Niveles de triglicéridos y desenlace de Pancreatitis aguda





Nawaz H et al. Am J Gastroenterol 2015;110:1497–1503.

Síndrome de quilomicronemia familiar

Desorden metabólico raro
1/1.000.000 a 1/250.00

→ Autosómico recesivo para el gene Lipoproteín-lipasa

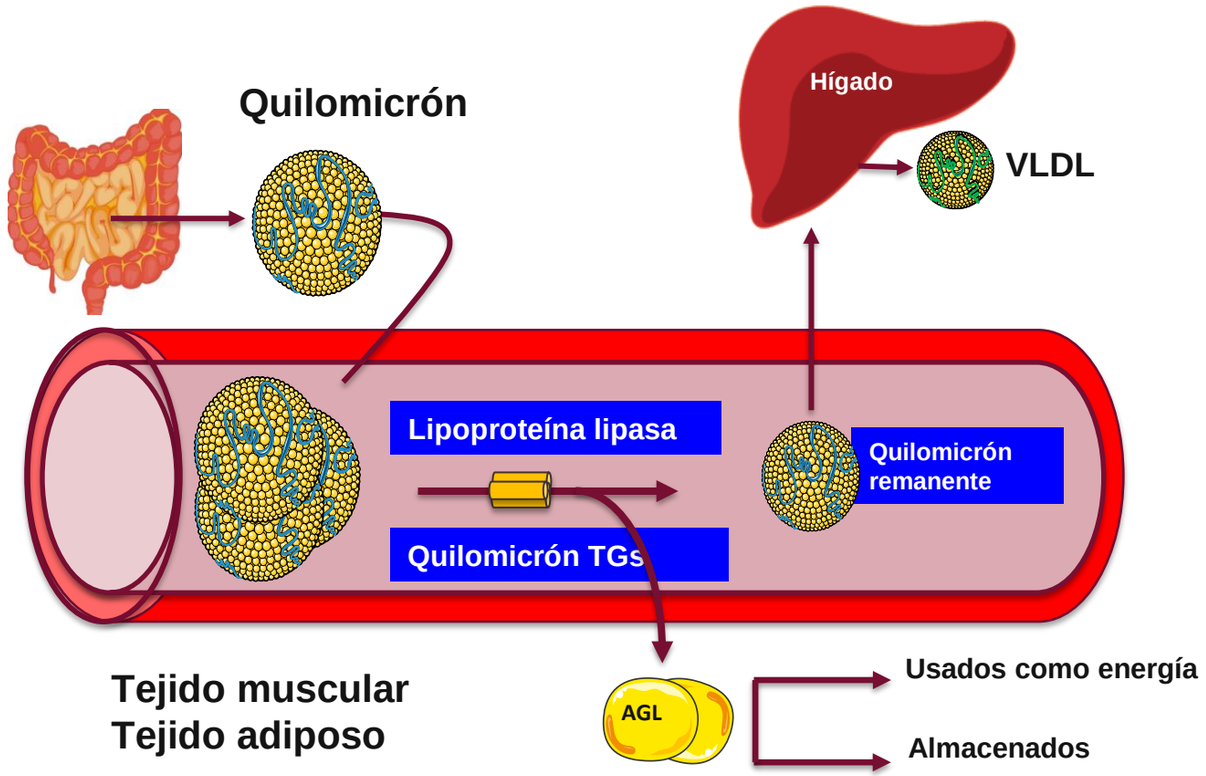
↓ Hidrólisis de lipoproteínas Ricas en triglicéridos

↓ Hiper quilomicronemia
Severa hipertrigliceridemia

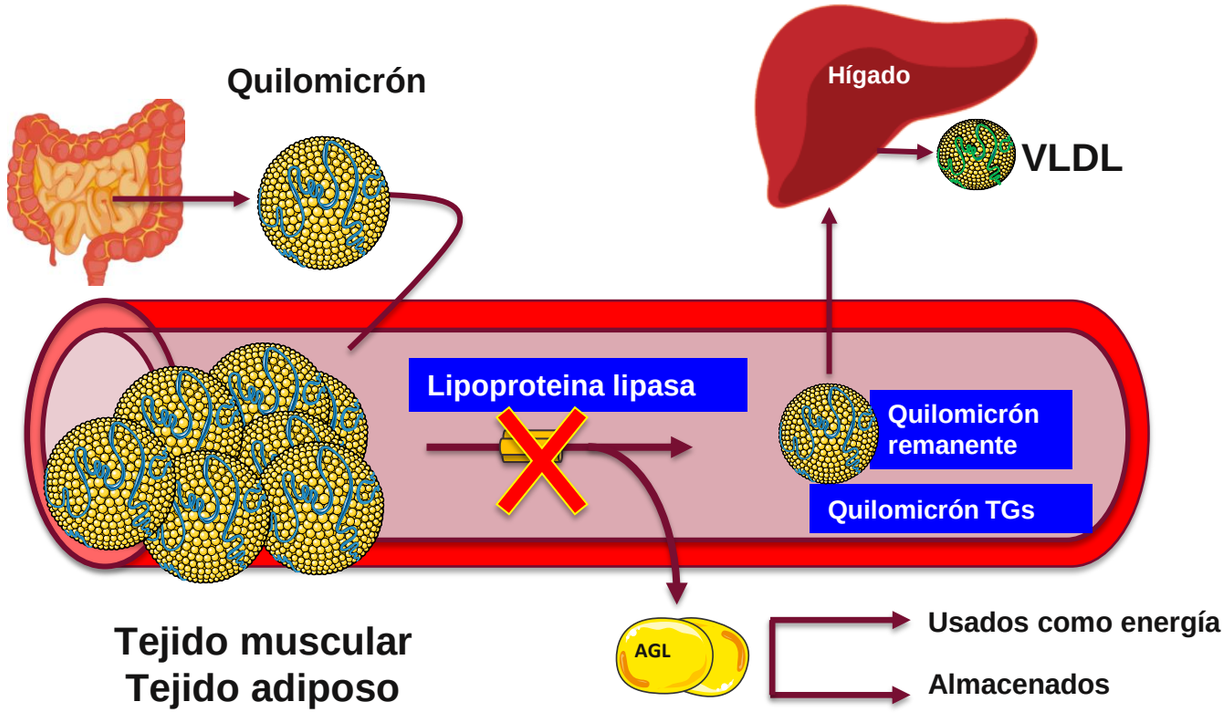
↓ **Deficiencia lipoprotein lipasa**

Brahm AJ, Nat Rev Endocrinol. 2015;11:352–362
Bass A, J Intern Med. 2020 ;287:340-8
Chyzhyk V, Trends Cardiovasc Med. 2020;30:80-85.

Sano



Quilomicronemia Familiar



Brunzell JDMed Clin North Am 1982;66:455–68; 2.
Brahm AJ. Nat Rev Endocrinol 2015;11:352–62

Síndrome de quilomicronemia familiar

Xantomas cutáneos
Tronco, nalgas extremidades

TG > 2000mg/dl

Lipemia retinalis

TG > 4000mg/dl

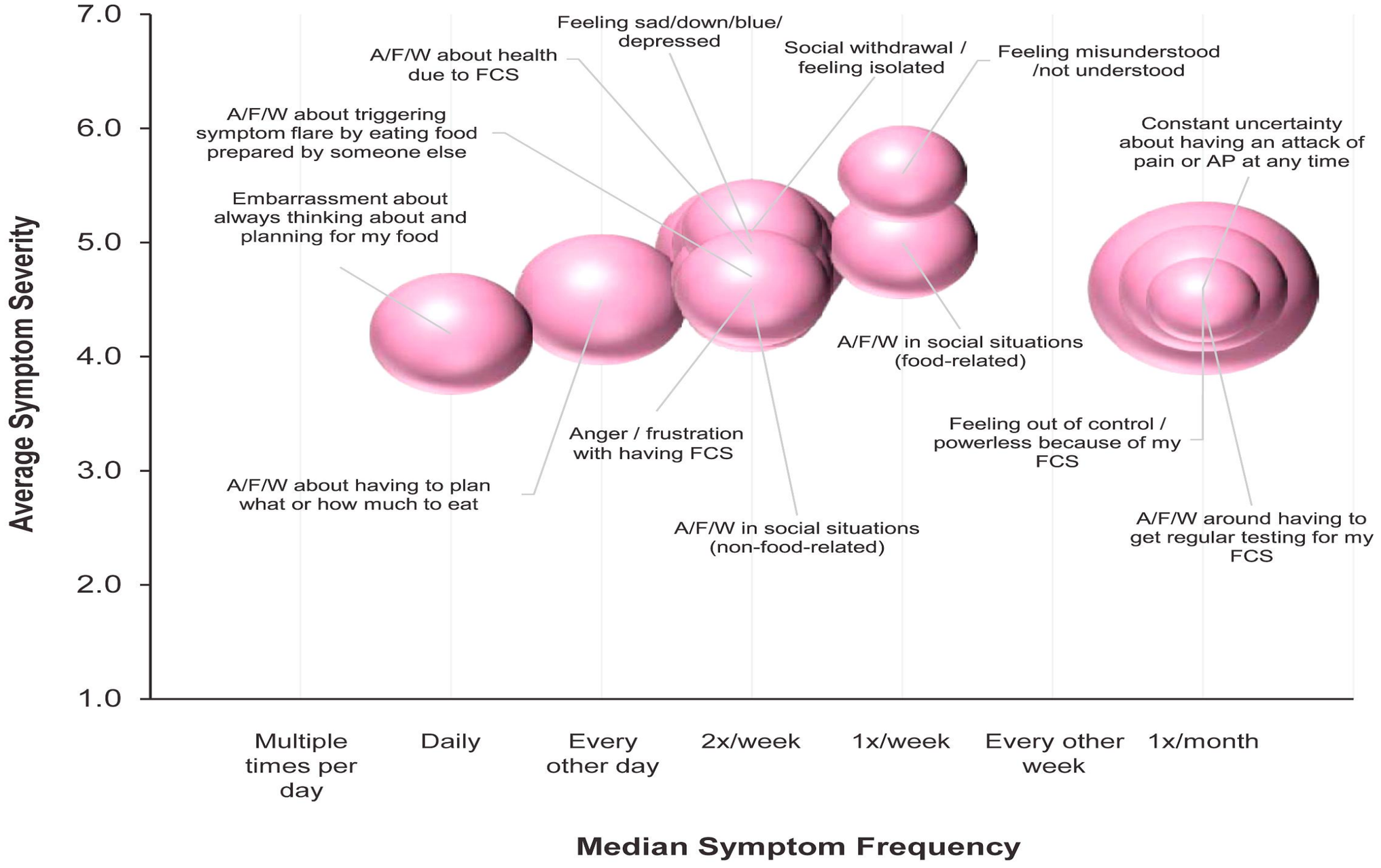
Bass A, J Intern Med. 2020 ;287:340-8
Kumar J, N Engl J Med 2005; 353:823
Chyzhyk V, Trends Cardiovasc Med. 2020;30:80-85.

Manifestations or complications of FCS	Prevalence
Eruptive xanthomas	17–23%
Lipaemia retinalis	4–36%
Hepatosplenomegaly or splenomegaly	12–25%
Abdominal pain	26–63%
Pancreatitis	60–88%
Multiple pancreatitis	17–48%

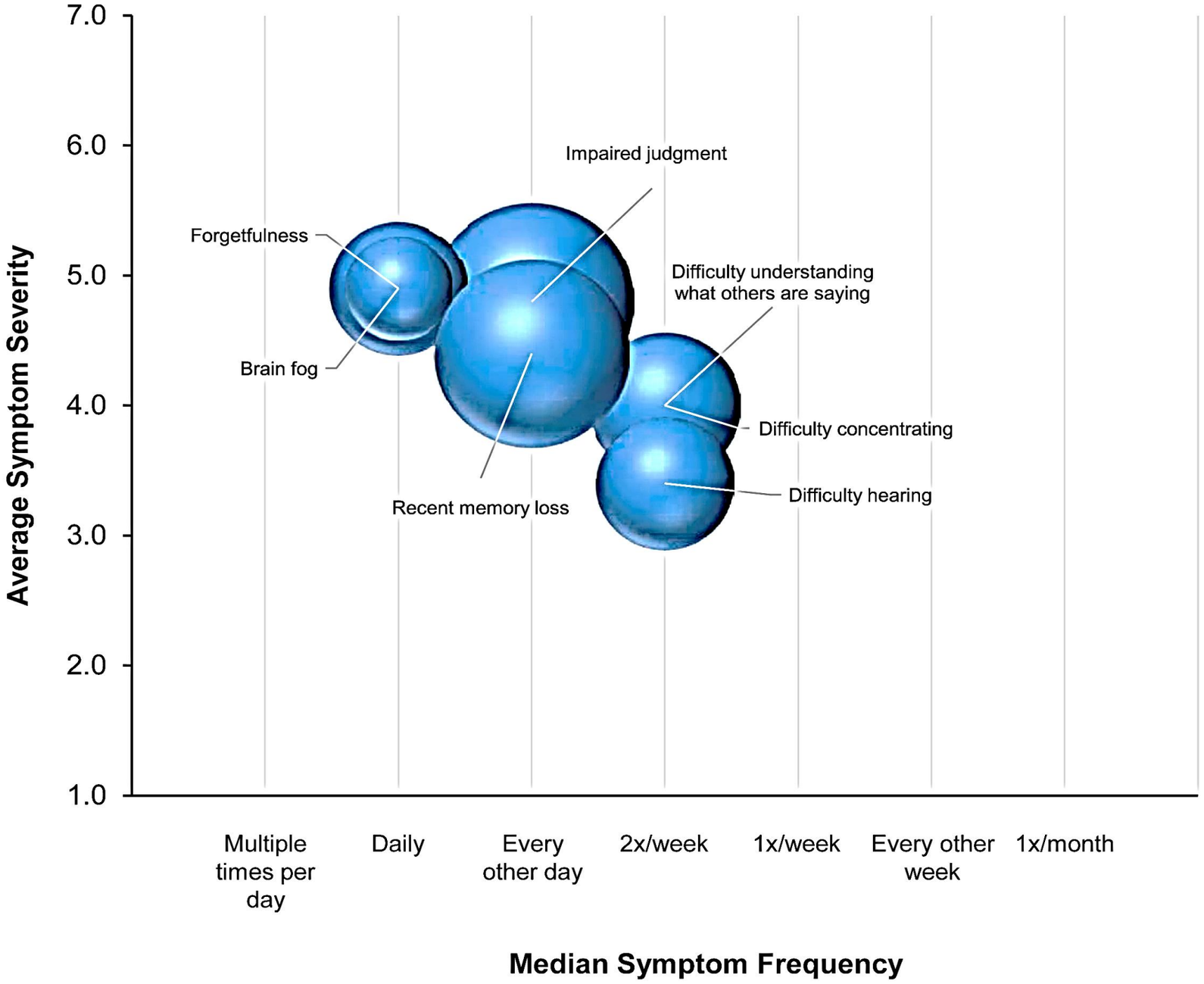
FCS, familial chylomicronemia syndrome.

Ansiedad y temor pancreatitis fatal
Dificultad adherirse dieta
Desórdenes alimentación >20%

Bass A, J Intern Med. 2020 ;287:340-8
Davidson M, J Clin Lipidol 2018; 12: 898–907
Gelrud A, et al. Expert Rev Cardiovasc Ther. 2017;15:879-87



Davidson M, et al. J Clin Lipidol. 2018;12:898-907



Davidson M, et al. J Clin Lipidol. 2018;12:898-907

The burden of familial chylomicronemia syndrome: interim results from the IN-FOCUS study

Michael Davidson[a], Michael Stevenson[b], Andrew Hsieh[b], Zahid Ahmad[c], Caroline Crowson[d] and Joseph L Witztum[e]

Top 7 symptoms in most severe form by domain	Severity[a]	Average reported frequency	Percent reporting symptom
Physical symptoms			
Bloating/distended stomach	4.7	Twice weekly	35
Generalized abdominal pain	4.6	Every other day	33
Feeling of physical weakness (i.e. asthenia)	4.8	Twice weekly	33
Fatigue	5.2	Every other day	27
Indigestion	4.3	Twice weekly	23
Pancreatic pain	6.2	Monthly	23
Lack of appetite	4.2	Twice weekly	22
Cognitive symptoms			
Difficulty concentrating	3.9	Every other day	18
'Brain fog'	4.7	Daily	17
Forgetfulness	5.8	Daily	10
Impaired judgment	4.8	Weekly	8
Recent memory loss	4.2	Every other day	8
Difficulty hearing	3.4	Daily	8
Difficulty understanding what others are saying	4.3	Twice weekly	5
Emotional Symptoms			
Constant uncertainty about having an attack of pain or acute pancreatitis at any time	5.2	Every other day	33
Anxiety/fear/worry about my health due to FCS	5.2	Twice weekly	30
Feeling out of control/powerless because of my FCS	4.9	Twice weekly	25
Anxiety/fear/worry about having to plan what to eat or how much I can eat	4.9	Every other day	22
Anxiety/fear/worry in social situations for nonfood reasons	4.5	Twice weekly	22
Feeling sad/down/blue/depressed	5.3	Daily	22
Social withdrawal/feeling isolated	5.3	Twice weekly	22

[a]Severity was recorded on a 1–7 Likert scale where 1 = mild and 7 = very severe.

Davidson M, et al. Expert Rev Cardiovasc Ther 2017;15:415–23.

The burden of familial chylomicronemia syndrome from the patients' perspective

Andres Gelrud[a], Karren R. Williams[b], Andrew Hsieh[b], Andrea R. Gwosdow[b], Alan Gilstrap[b] and Alan Brown[c]

[a]Center for Pancreatic Disorders, University of Chicago, Chicago, IL, USA; [b]Akcea Therapeutics Inc, A Subsidiary of Ionis Pharmaceuticals, Cambridge, MA, USA; [c]Division of Cardiology, Advocate Lutheran General Hospital, Park Ridge, IL, USA

Table 2. Demographics and characteristics of patients with FCS ($N = 10$).

Sex	Age (yrs)	Method of diagnosis	Age at onset of symptoms	Highest plasma TG value (mg/dL)	Average plasma TG value (mg/dL)
F	26	Genetic	7 yrs	3,000	700–800
M	36	Clinical	3 mo	4,800	1,200–1,800
M	37	Genetic	3 mo	2,600	500–700
M	46	Clinical	40 yrs	14,600	1,100
F	51	Genetic	20–25 yrs	4,400	1,200–1,800
F	57	Clinical	45 yrs	12,000	1,000
M	42	Clinical	8–10 yrs	27,000	1,000–2,000
M	50	Clinical	20–25 yrs	13,000	700–800
M	67	Genetic	49 yrs	9,000	2,600
F	55	Genetic	1.5 mo	26,000	3,100
Totals (Median)	48		15.5 yrs	10,500 mg/dL	1,300 mg/dL

Gelrud A, et al. Expert Rev Cardiovasc Ther. 2017;15:879-87.

The burden of familial chylomicronemia syndrome from the patients' perspective

Andres Gelrud[a], Karren R. Williams[b], Andrew Hsieh[b], Andrea R. Gwosdow[b], Alan Gilstrap[b] and Alan Brown[c]

[a]Center for Pancreatic Disorders, University of Chicago, Chicago, IL, USA; [b]Akcea Therapeutics Inc, A Subsidiary of Ionis Pharmaceuticals, Cambridge, MA, USA; [c]Division of Cardiology, Advocate Lutheran General Hospital, Park Ridge, IL, USA

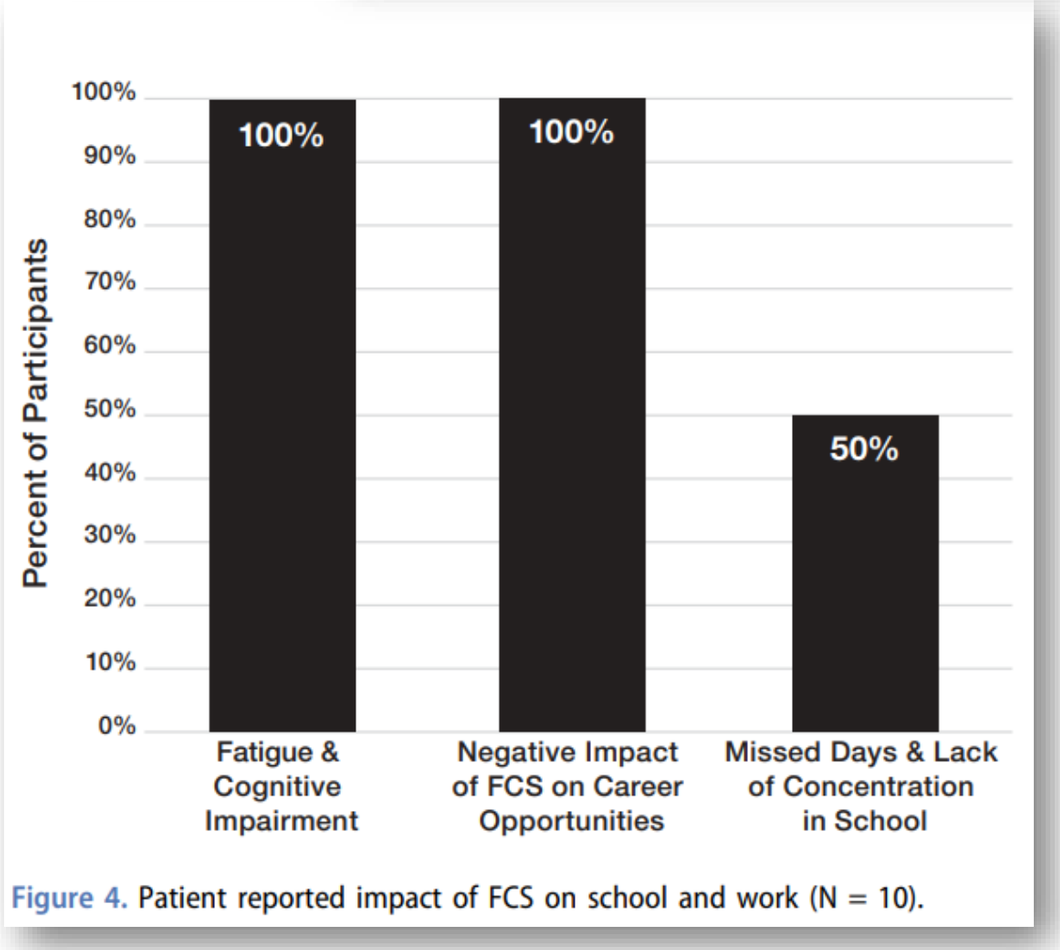
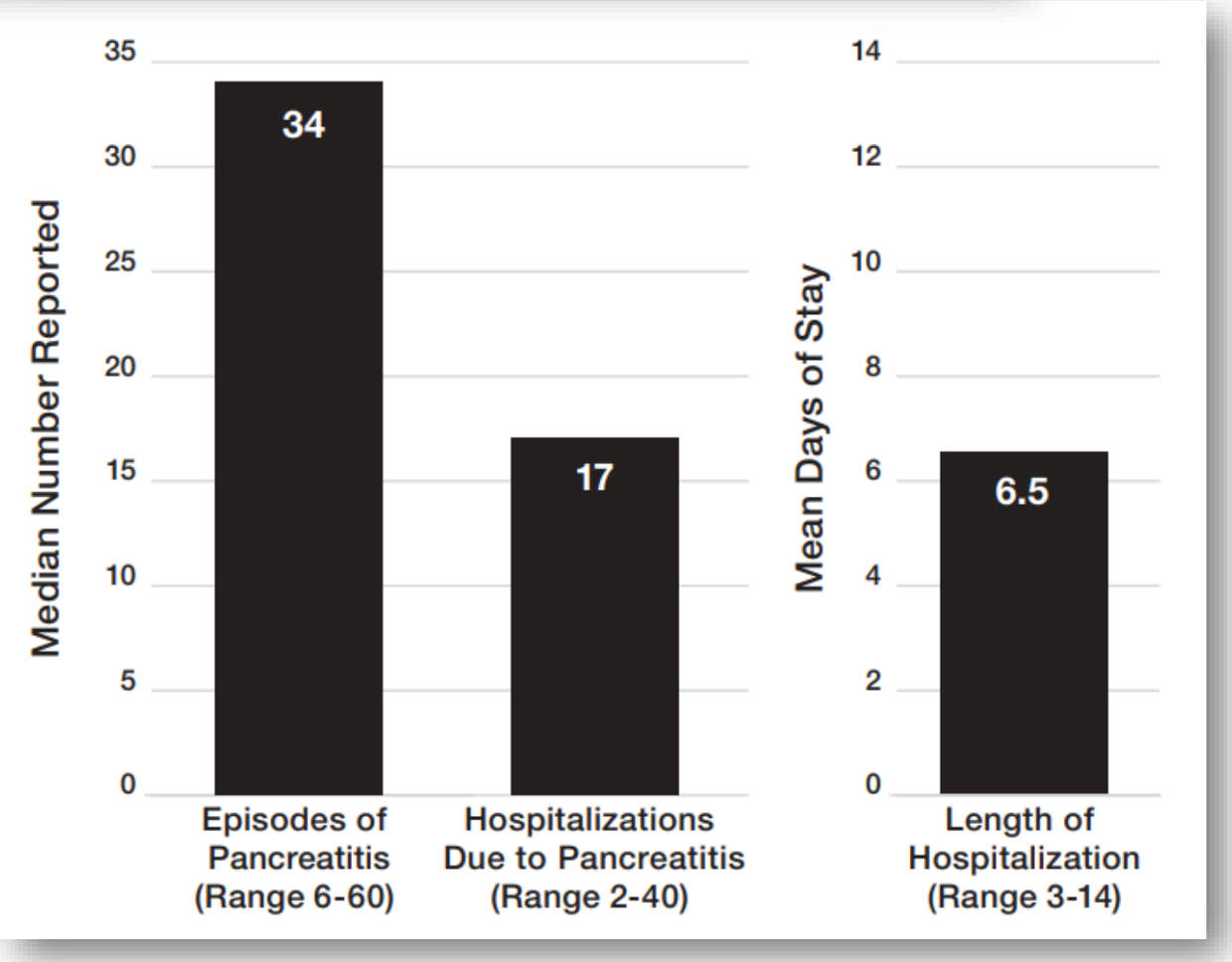
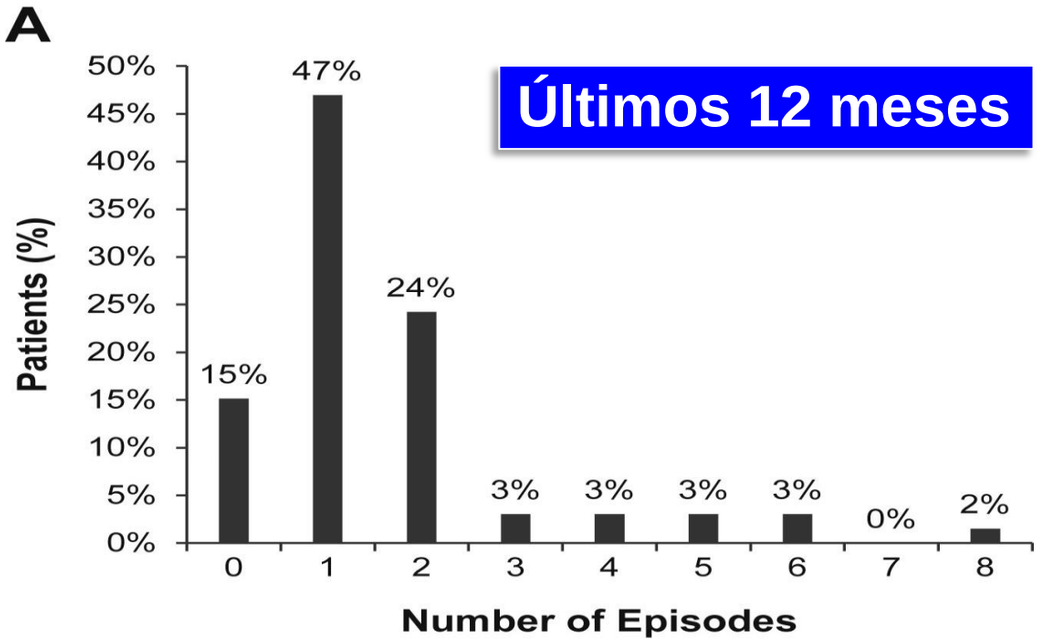


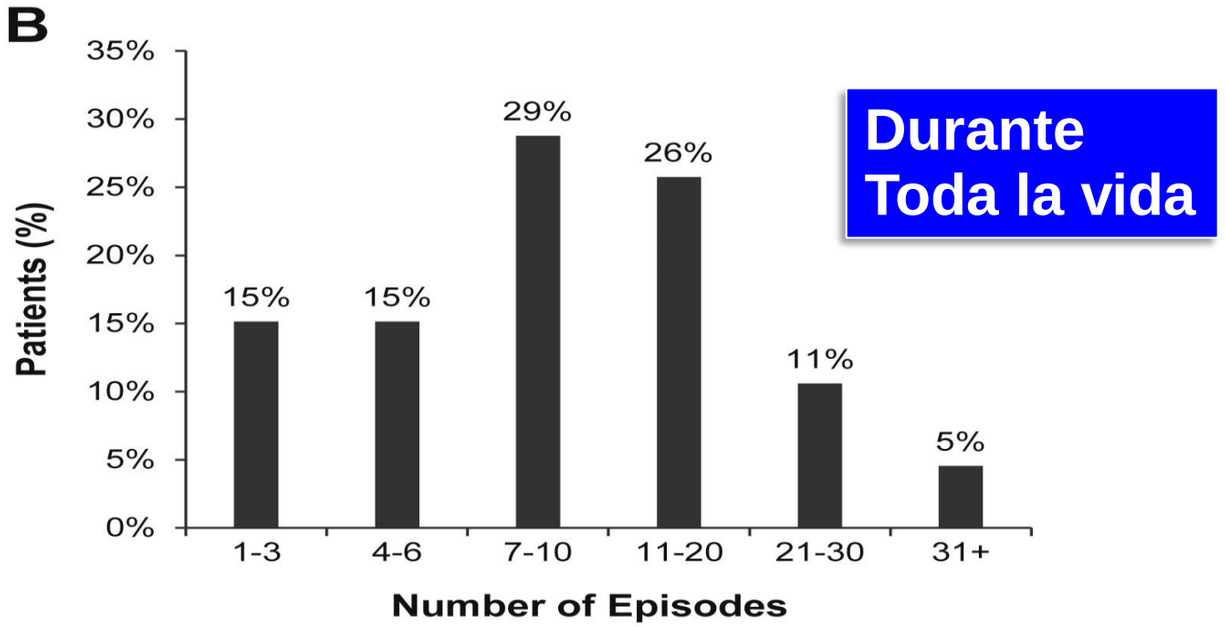
Figure 4. Patient reported impact of FCS on school and work (N = 10).



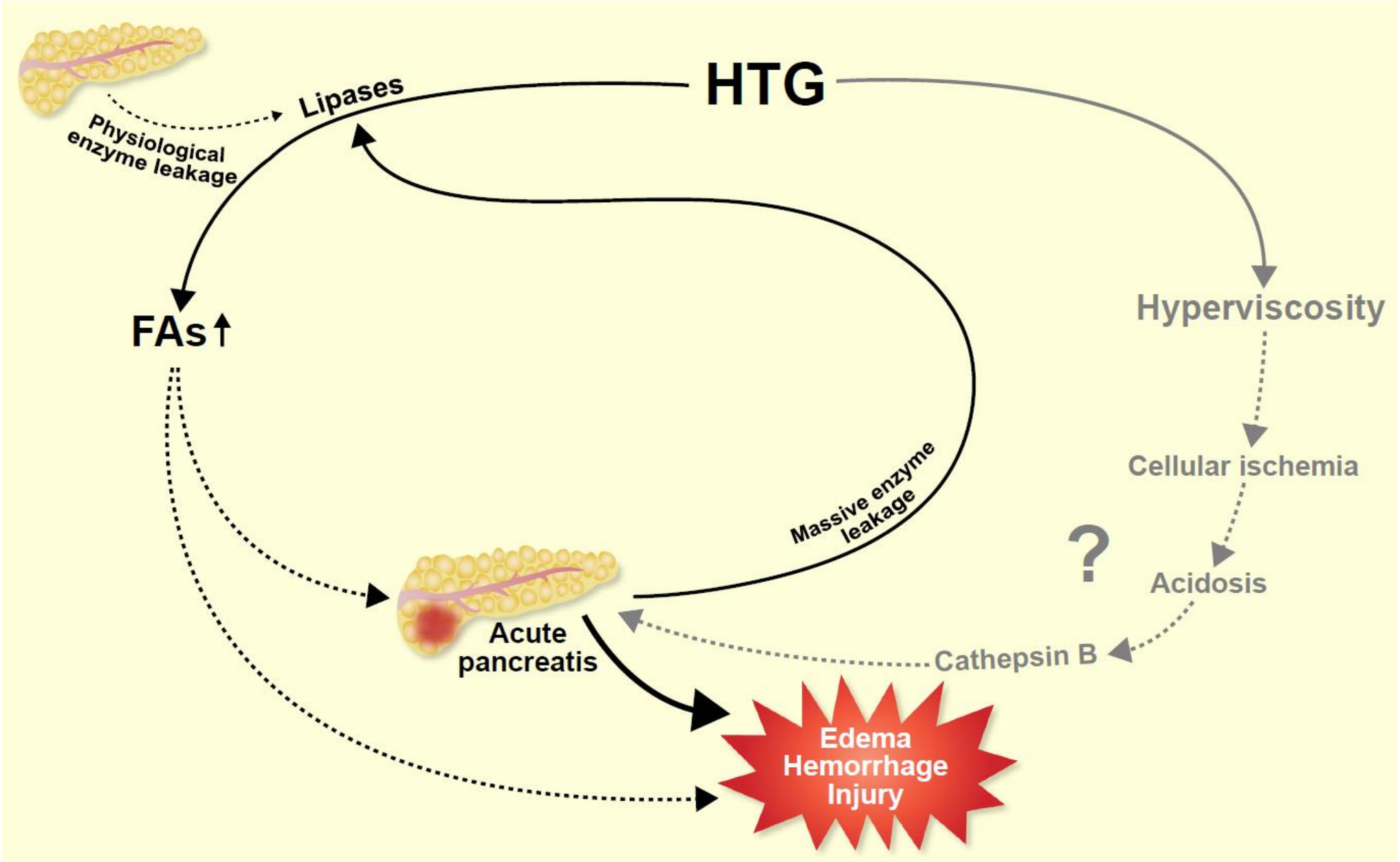
Gelrud A, et al. Expert Rev Cardiovasc Ther. 2017;15:879-87.

Episodios de Pancreatitis

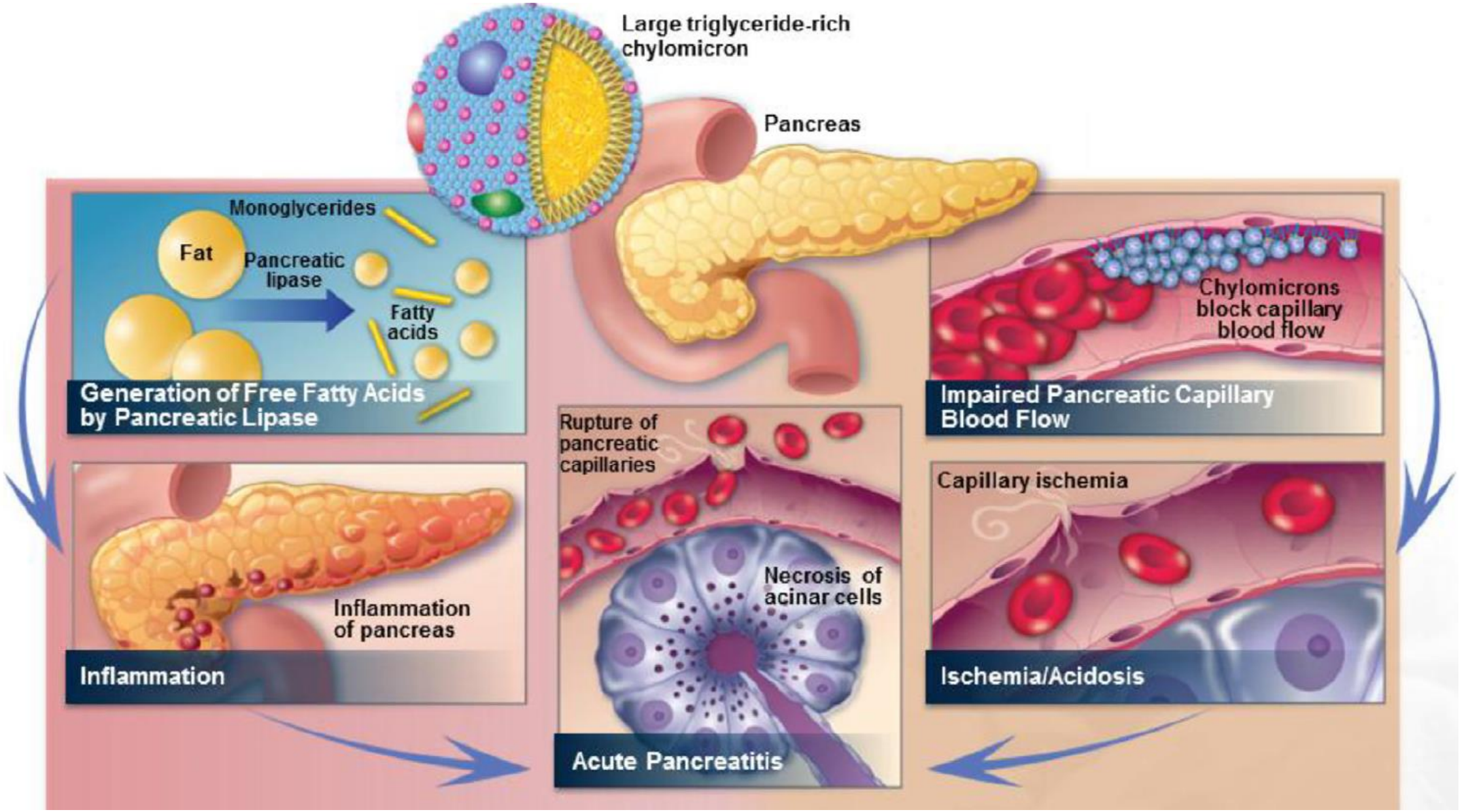




Davidson M, et al. J Clin Lipidol. 2018;12:898-907



Kiss L, et al. Act Physiol. 2023;237:e13916.



Chyzhyk V, Trends Cardiovasc Med. 2020;30:80-85

Quilomicronemia familiar, Diagnóstico

Hipertrigliceridemia severa, refractaria

TG en ayunas > 880 mg/dL

+

Mínima o no respuesta hipolipemiantes

Historia clínica

Pancreatitis aguda

o

Dolor abdominal Recurrente sin causas identificables

Ausencia causas Secundarias

Exceso de alcohol o
Diabetes incontrolada o
Medicamentos (BB, antisicoticos Esteroides, inhib. Proteasas) o
Enfermedades causen Hipertrigliceridemia
Cushing VIH IRC LES

Chyzhyk V, et al. Trends Cardiovasc Med. 2020;30:80-85

Review article

Identification and diagnosis of patients with familial chylomicronaemia syndrome (FCS): Expert panel recommendations and proposal of an “FCS score”

Philippe Moulin [a], Robert Dufour [b], Maurizio Averna [c], Marcello Arca [d], Angelo B. Cefalù [c], Davide Noto [c], Laura D'Erasmo [d], Alessia Di Costanzo [d], Christophe Marçais [a], Luis Antonio Alvarez-Sala Walther [e], Maciej Banach [f], Jan Borén [g], Robert Cramb [h], Ioanna Gouni-Berthold [i], Elizabeth Hughes [j], Colin Johnson [k], Xavier Pintó [l], Željko Reiner [m], Jeanine Roeters van Lennep [n], Handrean Soran [o], Claudia Stefanutti [p], Erik Stroes [q], Eric Bruckert [r, *]

Moulin P, et al. Atherosclerosis. 2018;275:265-272.

Hipertrigliceridemia severa refractaria

Severa = TGs en ayunas >10 mmol/L o 880 mg/dL

Puntaje para seleccionar los pacientes

Parámetro	Puntaje
TGs en ayunas >880 mg/dL (10 mmol/L) en 3 mediciones consecutivas de sangre	5[a]
TGs en ayunas >1760 mg/dL (20 mmol/L) al menos una vez	+1
Alguna medición previa de TGs < 175 mg/dL (2 mmol/L)	-5
Sin factor secundario[b] (excepto embarazo[c] y etinil estradiol)	2[d]
Antecedentes de pancreatitis	1
Dolor abodominal recurrente inexplicado	1
Sin antecedente de hiperlipidemia familiar combinada (HFC)	1
Falta de respuesta a la terapia hipolipemiante	1
Inicio de síntomas[e] a la edad: <40 años <20 años <10 años	 1 2 3

IMC <26 kg/m2
cLDL < 40 mg/dl

≥10 Muy probable SQF
≤9 Poco probable SQF
<8 Muy poco probable SQF

PTC Therapeutics

Moulin P, et al. Atherosclerosis. 2018;275:265-272.

The burden of familial chylomicronemia syndrome: interim results from the IN-FOCUS study

Michael Davidson[a], Michael Stevenson[b], Andrew Hsieh[b], Zahid Ahmad[c], Caroline Crowson[d] and Joseph L Witztum[e]

Path to Diagnosis	*N* (%)
Number of physicians seen for symptoms before FCS diagnosis, mean (median)	5 (5)
Physician specialty who made FCS diagnosis, *n* (%)	
I don't know	21 (35)
Endocrinologist	10 (17)
Pancreatologist	10 (17)
Lipidologist	6 (10)
Cardiologist	4 (7)
Prior to being correctly diagnosed with FCS	
Symptoms were misdiagnosed, *n* (%)	40 (67)
Most common misdiagnoses, *n* (%)	
Acute pancreatitis of unknown cause	19 (48)
Hypertriglyceridemia	18 (45)
Gallstones	6 (15)

Davidson M, et al. Expert Rev Cardiovasc Ther 2017;15:415–23.

Quilomicronemia familiar
Tratamiento

Incapacidad de metabolizar Quilomicrones

Restricción de grasa en la dieta
30-50 gr/d 0 15-25% del total energía
Difícil adherencia

Chyzhyk V, Trends Cardiovasc Med. 2020;30:80-85

The NEW ENGLAND JOURNAL of MEDICINE

ORIGINAL ARTICLE

Volanesorsen and Triglyceride Levels in Familial Chylomicronemia Syndrome

J.L. Witztum, D. Gaudet, S.D. Freedman, V.J. Alexander, A. Digenio, K.R. Williams, Q. Yang, S.G. Hughes, R.S. Geary, M. Arca, E.S.G. Stroes, J. Bergeron, H. Soran, F. Civeira, L. Hemphill, S. Tsimikas, D.J. Blom, L. O'Dea, and E. Bruckert

N Engl J Med 2019;381:531-42.

Drugs (2019) 79:1349–1354
https://doi.org/10.1007/s40265-019-01168-z

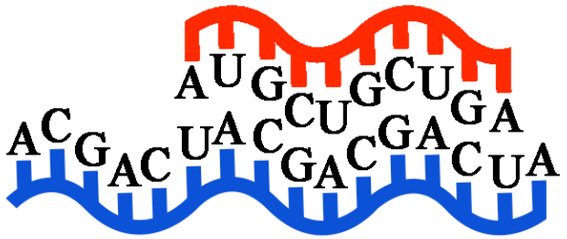
ADISINSIGHT REPORT

Volanesorsen: First Global Approval

Julia Paik[1] · Sean Duggan[1]

Drugs 2019; 79:1349-54

Oligonucleótido antisentido



ARN mensajero

Bloquea APO CIII

<< Triglicéridos 56-76%

Drug Design, Development and Therapy

Dovepress
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REVIEW

Volanesorsen in the Treatment of Familial Chylomicronemia Syndrome or Hypertriglyceridaemia: Design, Development and Place in Therapy

This article was published in the following Dove Press journal:
Drug Design, Development and Therapy

Oluwayemisi Esan
Anthony S Wierzbicki

Drug Design, Development and Therapy 2020:14 2623–36

The NEW ENGLAND JOURNAL of MEDICINE

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Volanesorsen and Triglyceride Levels in Familial Chylomicronemia Syndrome

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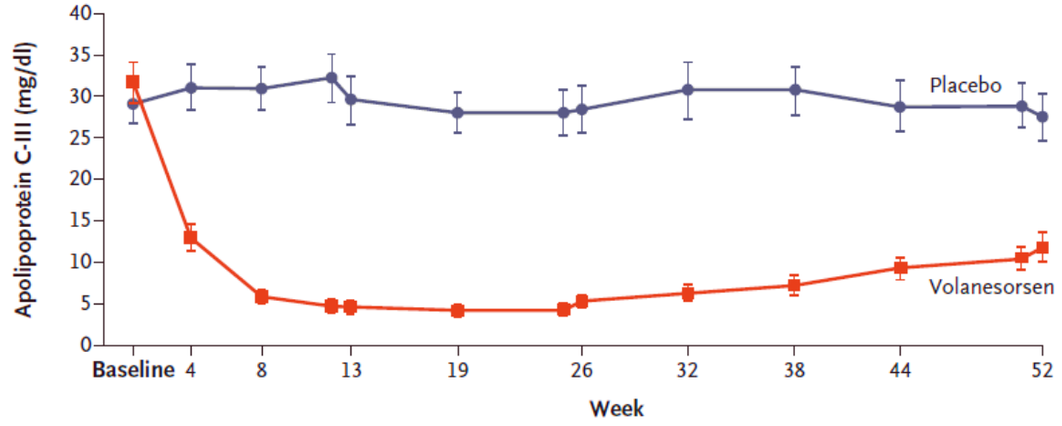
Witztum JL, N Engl J Med 2019;381:531-42.

Table 1. Characteristics of the Patients at Baseline.*

Characteristic	Placebo (N=33)	Volanesorsen (N=33)	All Patients (N=66)
Mean age (range) — yr	46 (20–68)	47 (22–75)	46 (20–75)
Sex — no.			
Female	19	17	36
Male	14	16	30
Body-mass index†	24.1±4.7	25.9±6.5	25.0±5.7
Triglycerides — mg/dl	2152	2267	2209
History of pancreatitis — no. (%)	26 (79)	24 (73)	50 (76)
Baseline use of n–3 fatty acids, fibrates, or both — no. (%)	16 (48)	19 (58)	35 (53)
Genetic mutations — no. (%)			
LPL	24 (73)	17 (52)	41 (62)
APOA5	1 (3)	1 (3)	2 (3)
GPIHBP1	0	5 (15)	5 (8)
LMF1	0	1 (3)	1 (2)
APOC2	0	1 (3)	1 (2)
LPL/LMF-1	0	1 (3)	1 (2)
LPL/APOA5	1 (3)	0	1 (2)
Not identified‡	7 (21)	7 (21)	14 (21)

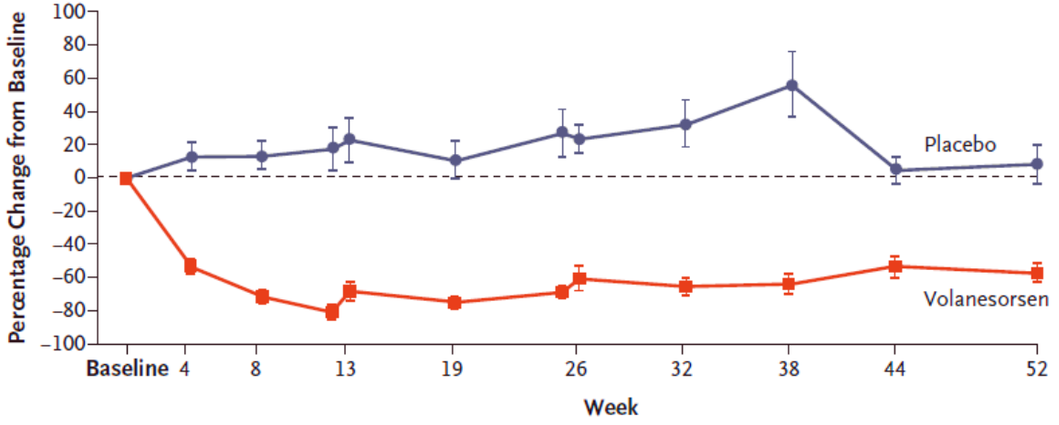
Witztum JL, N Engl J Med 2019;381:531-42.

A Change in Fasting Apolipoprotein C-III Levels over Time



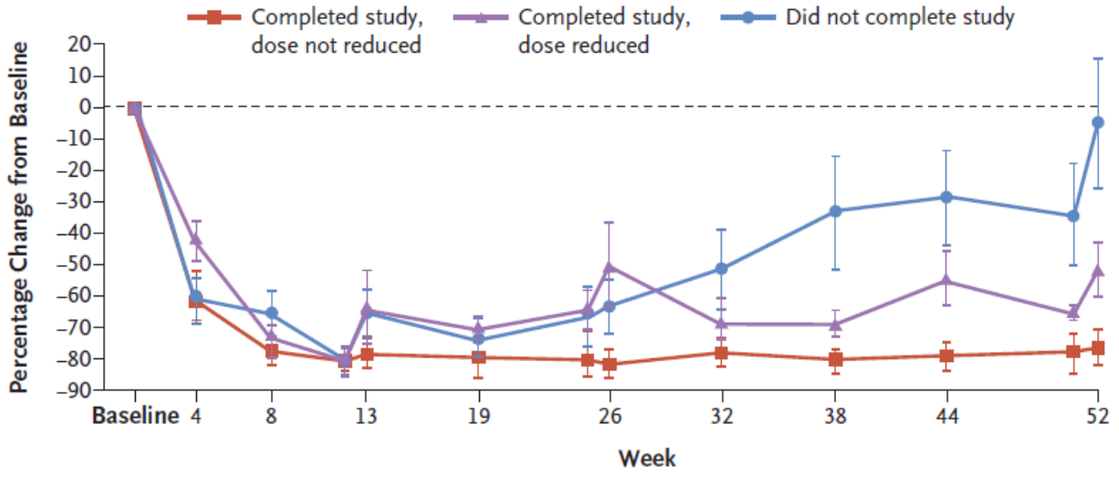
No. at Risk										
Placebo	33	31	33	26 32	32	26 30	31	29	30	26 26
Volanesorsen	33	30	33	28 30	28	22 27	25	24	25	23 24

B Change in Triglyceride Levels over Time



No. at Risk									
Placebo	31	33	26 32	31	26 30	31	29	30	26
Volanesorsen	30	33	28 30	28	22 27	25	24	25	24

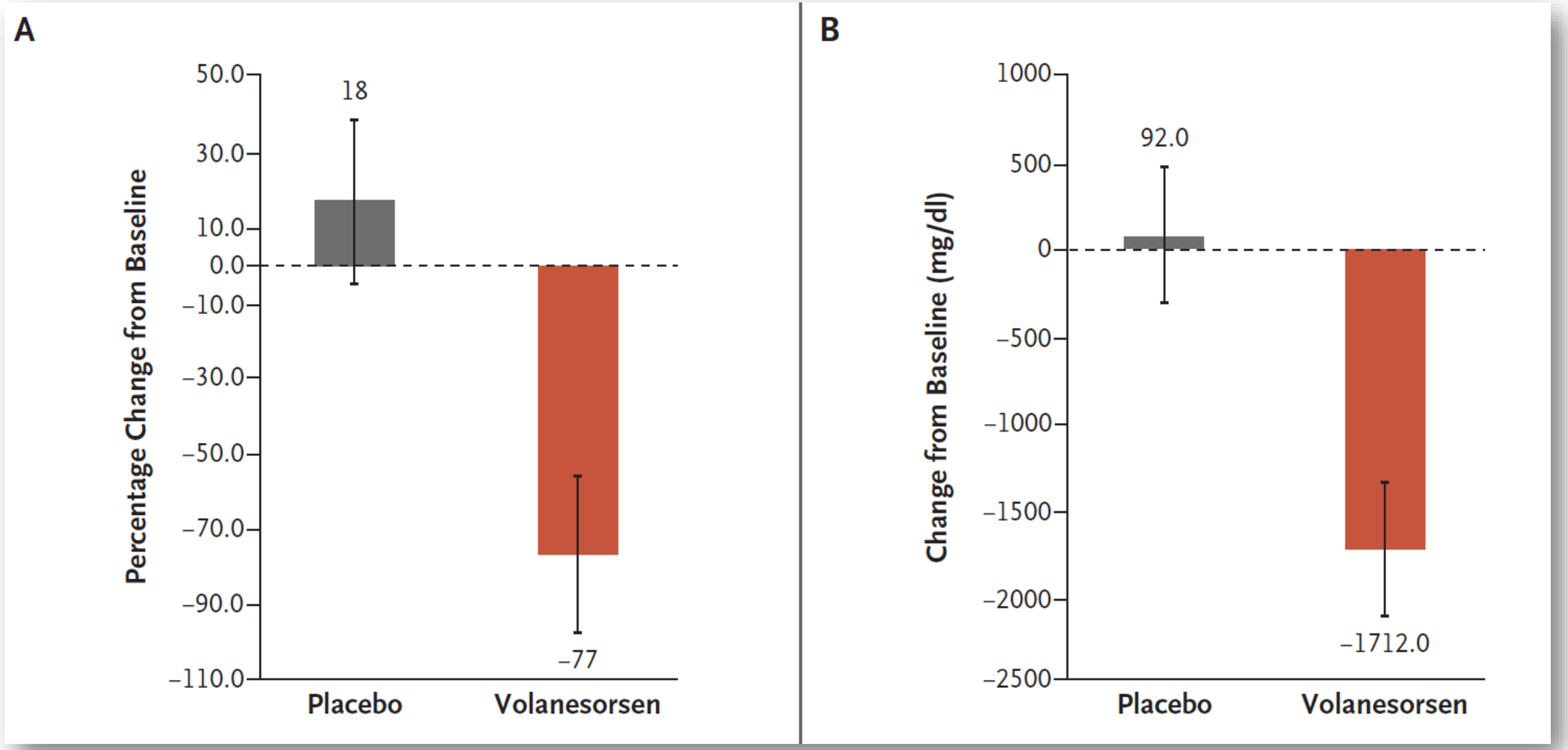
C Change in Triglyceride Levels over Time with and without Volanesorsen Dose Adjustment



No. at Risk										
Completed study, dose not reduced	6	5	6	6 6	6	5 5	5	6	6	4 6
Completed study, dose reduced	13	13	13	12 12	13	11 13	13	13	13	12 12
Did not complete study	14	12	14	10 12	9	6 9	7	5	6	7 6

Witztum JL, N Engl J Med 2019;381:531-42.

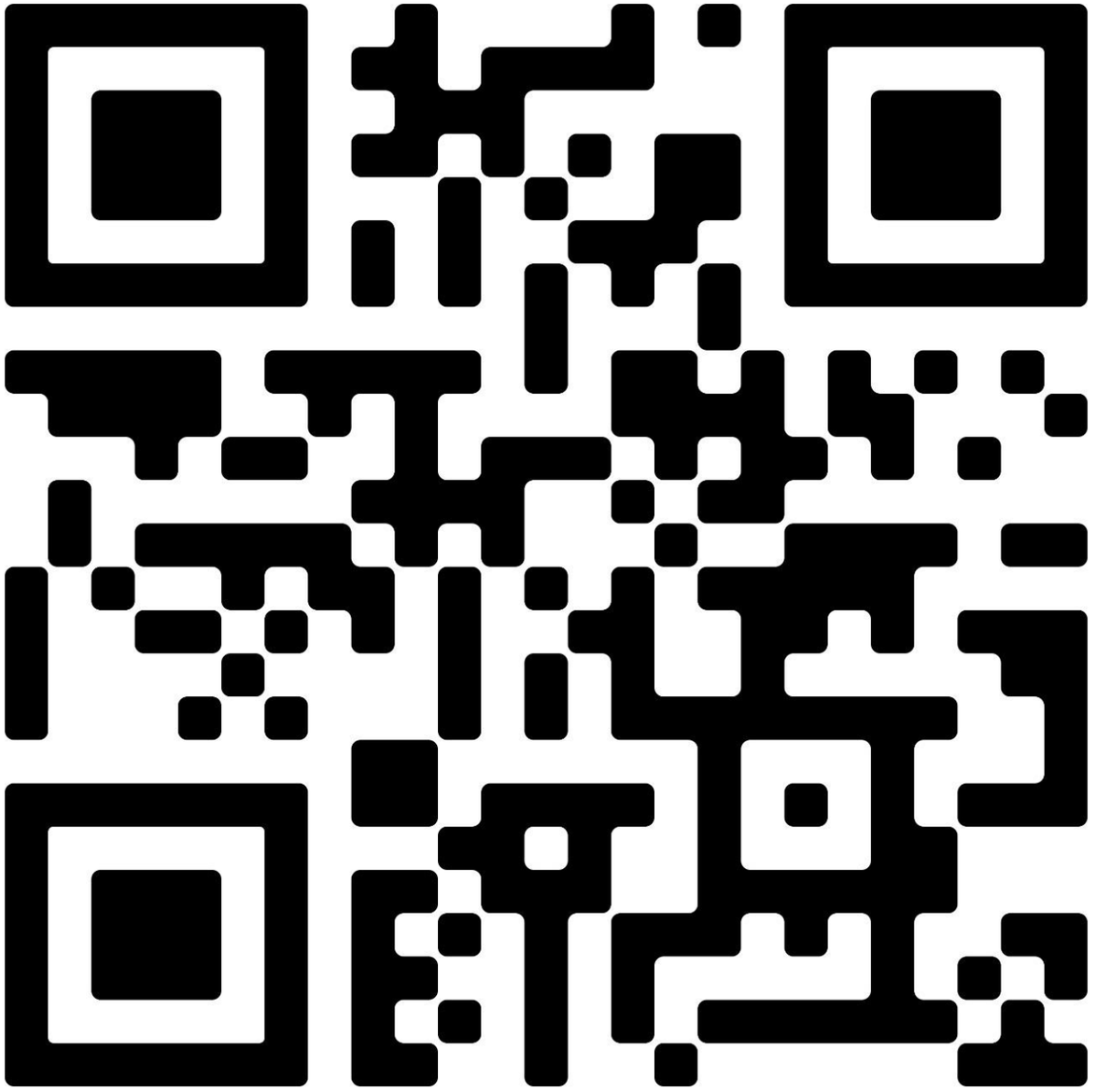
Cambio de triglicéridos en tres meses



Witztum JL, N Engl J Med 2019;381:531-42.

Mensajes para la casa

NAFLD/MAFLD subdiagnosticada
Alta mortalidad cardiovascular
Principal causa de cirrosis
Páncreas graso pancreatitis aguda crónica cáncer
Quilomicronemia familiar enfermedad huérfana
Altera severamente calidad de vida
Sospecharla pancreatitis recurrente >> triglicéridos



Muchas gracias!

1. Fasting TGs >10 mmol/L for 3 consecutive blood analyses (+5)[a]
   - Fasting TGs >20 mmol/L at least once (+1)
2. Previous TGs <2 mmol/L (-5)
3. No secondary factor[b] (except pregnancy[c] and ethinylestradiol) (+2)
4. History of pancreatitis (+1)
5. Unexplained recurrent abdominal pain (+1)
6. No history of familial combined hyperlipidaemia (+1)
7. No response (TG decrease <20%) to hypolipidaemic treatment (+1)
8. Onset of symptoms at age:
   - <40 years (+1)
   - <20 years (+2)
   - <10 years (+3)

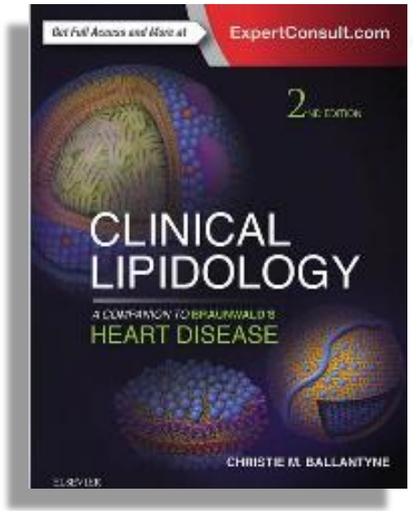
>880 mg

FCS score:
≥10: FCS very likely
≤9: FCS unlikely
≤8: FCS very unlikely

Moulin P, et al. Atherosclerosis. 2018;275:265-272.

Prevalencia de FCS

Prevalencia estimada citada por la literatura y las organizaciones...



1:1.000.000[1]

NORD®
National Organization for Rare Disorders

1:250.000[3]

1. Kastelein JP. NORD. (2015) https://rarediseases.org/physician-guide/lipoprotein-lipase-deficiency-lpld/. 2. Ballantyne, C. et al. Clinical Lipidology:A Companion to Braunwald's Heart Disease. 2nd ed. Elsevier;Saunders; 2014. 3. Kastelein JP. NORD. (2016) https://rarediseases.org/physician-guide/familial-lipoprotein-lipase-deficiency/. 3. https://pro.aace.com/sites/default/files/2019-02/FCS_082318_formatted.pdf

The burden of familial chylomicronemia syndrome from the patients' perspective

Andres Gelrud[a], Karren R. Williams[b], Andrew Hsieh[b], Andrea R. Gwosdow[b], Alan Gilstrap[b] and Alan Brown[c]

[a]Center for Pancreatic Disorders, University of Chicago, Chicago, IL, USA; [b]Akcea Therapeutics Inc, A Subsidiary of Ionis Pharmaceuticals, Cambridge, MA, USA; [c]Division of Cardiology, Advocate Lutheran General Hospital, Park Ridge, IL, USA

Gelrud A, et al. Expert Rev Cardiovasc Ther. 2017;15:879-87.

Table 3. Factors contributing to a reduced quality of life in patients with FCS.

Effect of low-fat diet

- Compliance with the diet is difficult; particularly when not at home
- Diet affects other members of the household and limits socialization
- Satisfaction with the diet is low
- Patients are not convinced that the diet is beneficial for reducing symptoms
- Financial cost of purchasing fat-free foods
- Burden of time required to research fat-free options and prepare fat-free meals

Effect of FCS on jobs and careers

- Fatigue and inability to concentrate due to pain can limit performance
- Frequent job absences can impede promotion
- Concerns about diet limit the ability to travel on the job
- Need for healthcare and insurance limits job opportunities
- Symptoms of FCS limit ability of patients to train for or perform work in a preferred career

Effect of FCS on social life

- Fatigue and dietary lifestyle considerations associated with FCS limit social life
- Failure of friends and family to understand the seriousness of FCS is a major irritant
- Some caregivers have difficulty adjusting to a reduced social life