“Enfermedad celíaca”

XII ENCUENTRO NACIONAL DE FAMILIAS

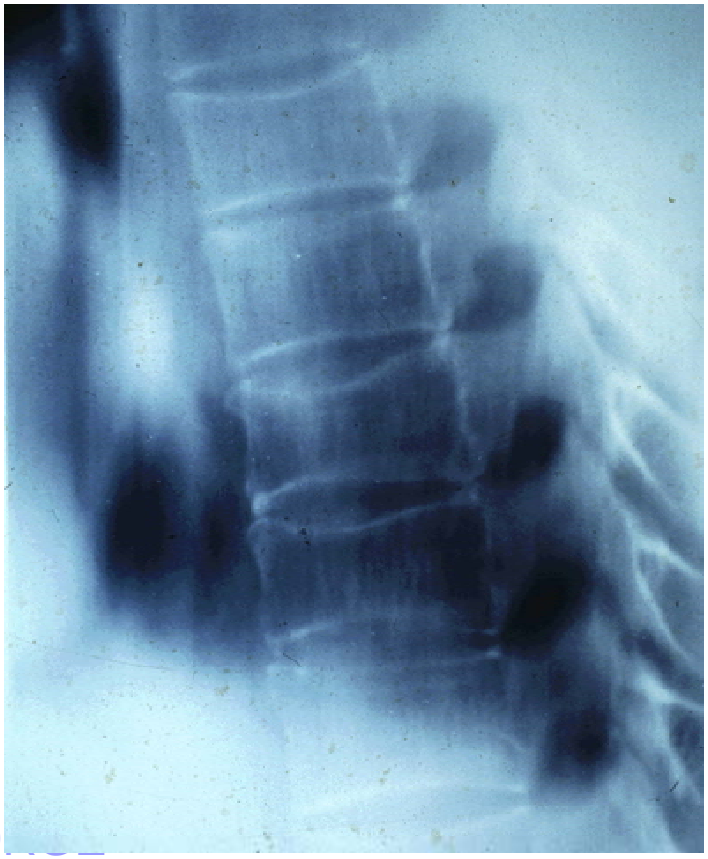
3 de noviembre de 2012

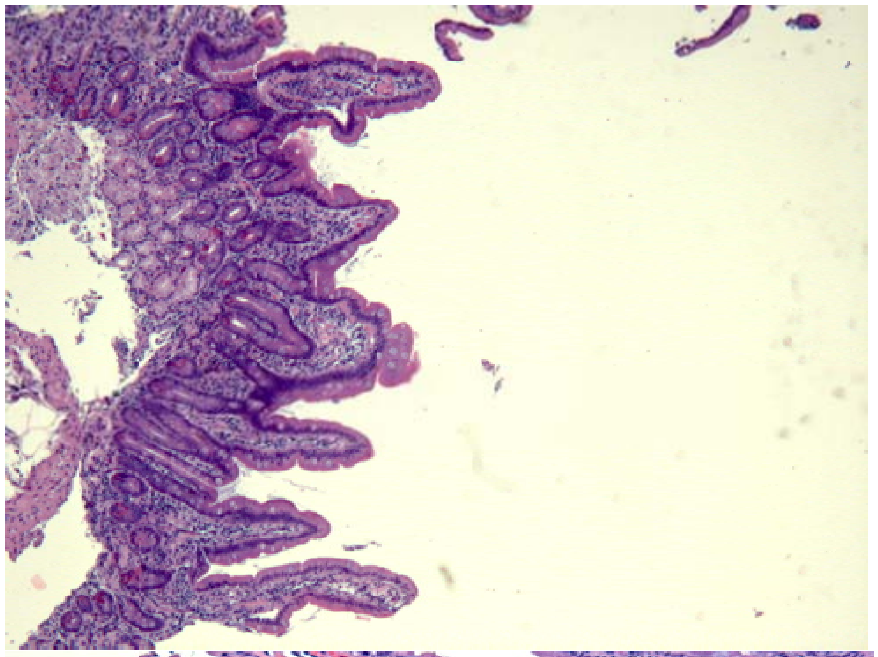
Dr. Miguel Montoro
Unidad de Gastroenterología y Hepatología
Hospital San Jorge Huesca

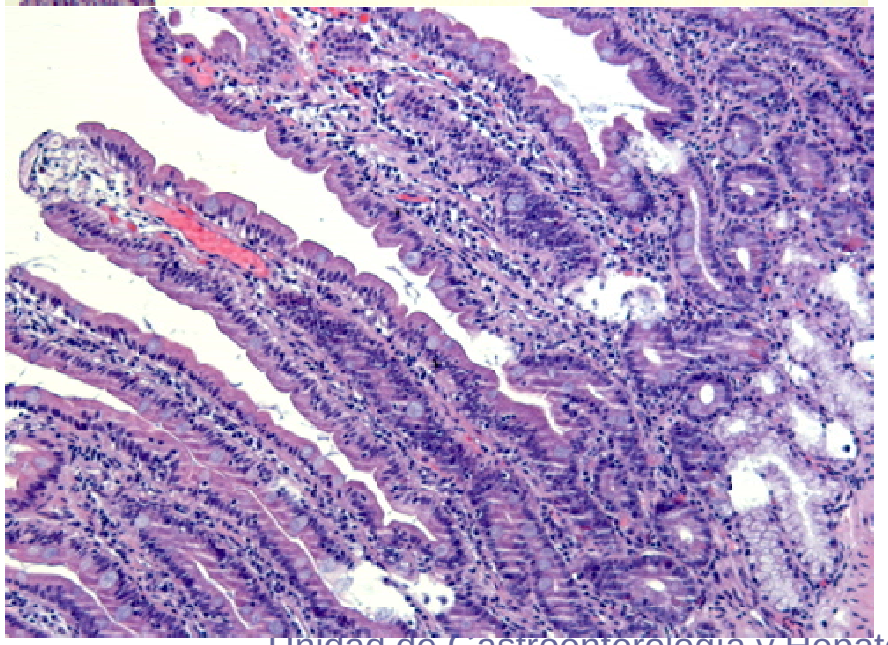
M.M.H. 34 años

Aplastamiento de D10

Osteoporosis idiopática



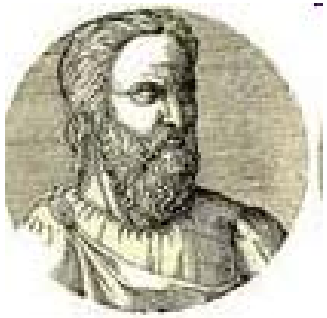




Francis Adams, médico escocés, nacido de una familia humilde. Trabajó como médico rural en Escocia durante muchos años y se dedicó a traducir al inglés obras clásicas de medicina de la antigua Grecia.

Banchory (Scotland), 1853

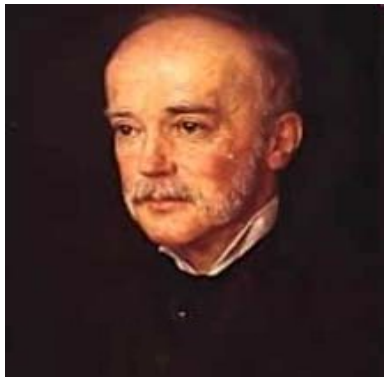
The Extant Works of Aretseus the Cappadocian [Las obras conservadas de Areteo de Capadocia] (1853), en una edición que incluía también el texto griego original.



Los trabajos de Aretaeus fueron editados y traducidos por Francis Adams e impresos por la Sydenham Society en 1856

"Si el vientre no realiza la digestión de los alimentos, la alimentación fluye sin digerir, inmutable y cruda, y no se transmite nada al conjunto del cuerpo, a tales cosas las denominamos celíaca…"

[Areteo de Capadocia, *Obra Médica*, L. VIII, 7 (1)]



Samuel Gee

Samuel Gee gave the first modern-day description of coeliac disease in a lecture at the Hospital for Sick Children, Great Ormond Street in 1887.

, pero no fue hasta 1888 cuando Samuel Gee daba a conocer un informe clínico claro sobre la condición celíaca con el título de “La afección celiaca”. En las explicaciones de Gee se incluyen la importancia de la alimentación en el tratamiento, que la proporción de farináceos debe ser mínima, y cómo la cura pasa por la dieta.

Guerra, hambruna, serendipia y epigenética

6 MARZO, 2010

by A. Aledo

tags: celiaquía, desnutrición, embarazo, epigenética, Genética, hambruna holandesa, Medicina, serendipia

Durante el último año de la II Guerra Mundial, Holanda vivió una de las mayores hambrunas ocurridas en un país moderno y desarrollado. Este acontecimiento ha servido como ejemplo científico de los efectos de la desnutrición a corto y largo plazo en una población, y en sus generaciones posteriores. Más aún, la escasez de algunos productos específicos condujo a un gran descubrimiento: el problema causante de la enfermedad celiaca.

Hagamos una foto escueta del acontecimiento histórico. Junio de 1944, las tropas aliadas penetran a través de la costa francesa en el llamado Desembarco de Normandía, que supone el principio del fin de la II Guerra Mundial en Europa. En agosto del mismo año, los Aliados consiguen liberar el sur de los Países Bajos pero, tras algunas revueltas en el sur del país, y el fracaso de la **Operación Market Garden** -destinada a apoderarse de los grandes puentes sobre el río Rin-, la administración militar alemana decreta el embargo sobre el transporte de comida a la zona que aún queda ocupada. Ésta fue la chispa de una concatenación de eventos que llevó al denominado por los holandeses como *Hongerwinter* (invierno del hambre). Así, las raciones diarias adultas en ciudades como Amsterdam o Rotterdam fueron de menos de 1000 Kcal a finales de 1944, y de incluso 580 Kcal diarias en las zonas costeras en febrero de 1945. La llamada Operación Maná puso fin a esta trágica etapa con el reparto de comida por parte de las fuerzas aéreas británicas en la zona aún ocupada al final de la guerra.

Sin embargo, la Hambruna Holandesa de 1944 es un ejemplo de una serie de terribles acontecimientos

Teclea y presiona ente

COMENTARIOS RECIENTES

Mike MC on Predica padre... que por u...

Elena on Predica padre... que por u...

Mike MC on Predica padre... que por u...

José Luis Contreras ... on Predica padre... que por u...

Mike MC on Detrás de toda gran persona ha...

Aurea Curiositas on Detrás de toda gran persona ha...

Mike MC on Detrás de toda gran persona ha...

Mike MC on El Colegio de Médicos de Murci...

ACREDITACIONES

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BIBLIOTECA DE ARTÍCULOS

país, y el fracaso de la **Operación Market Garden** -destinada a apoderarse de los grandes puentes sobre el río Rin-, la administración militar alemana decreta el embargo sobre el transporte de comida a la zona que aún queda ocupada. Ésta fue la chispa de una concatenación de eventos que llevó al denominado por los holandeses como *Hongerwinter* (invierno del hambre). Así, las raciones diarias adultas en ciudades como Amsterdam o Rotterdam fueron de menos de 1000 Kcal a finales de 1944, y de incluso 580 Kcal diarias en las zonas costeras en febrero de 1945. La llamada Operación Maná puso fin a esta trágica etapa con el reparto de comida por parte de las fuerzas aéreas británicas en la zona aún ocupada al final de la guerra.

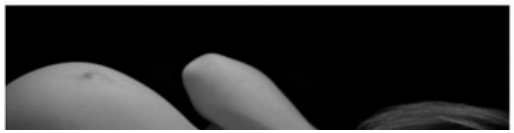
Sin embargo, la Hambruna Holandesa de 1944 es un ejemplo de una serie de terribles acontecimientos de la historia que han dejado un gran legado científico. Veamos por qué.

Serendipia en estado puro…

Fig. 1- La serendipia aparece cuando mentes preclaras hacen grandes descubrimientos en situaciones que resultan accidentes banales para los demás.

Según wikipedia, se define **serendipia** como **un descubrimiento o un hallazgo afortunado y sobre todo inesperado**, por accidente, y sobre todo gracias a la sagacidad del observador o investigador implicado. Es el modo por el cual Fleming descubrió la penicilina, y también el método por el cual se resuelven la mayoría de los casos en la serie **House MD**. Pero también fue la manera por la cual se descubrió la relación del trigo y el gluten con la **enfermedad celiaca**. El pediatra holandés Willen-Karel Dicke observó que algunos niños afectados por la enfermedad mejoraron de manera espectacular de su padecimiento con la escasez de productos derivados del trigo durante el *Hongerwinter*. Además, tras el fin de la ocupación alemana y el periodo de hambruna, los mismos niños volvieron a empeorar sorprendentemente. La brillante deducción de Dicke le llevó a diseñar en los años 50 la primera dieta libre de trigo para niños celiacos, la cual probó su hipótesis, cambió radicalmente la vida de estos enfermos, y supuso el primer paso para el descubrimiento del gluten y la gliadina en el trigo y otros productos.

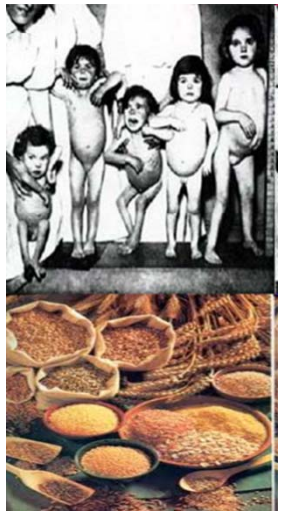
Una evidencia temprana de la epigenética.



Pero el gran legado dejado por la Hambruna Holandesa de 1944 fue la posibilidad del estudio a posteriori de la población superviviente sujeta a una fuerte desnutrición temporal. Así, aquellas

“ Dicke…el pediatra holandés”

- **. El pediatra holandés Dicke relata en tu tesis doctoral en 1950 cómo los niños celíacos mejoraban cuando se excluía de su dieta el trigo, el centeno y las harinas de avena. Si estos alimentos se sustituían por almidón de trigo, harina de maíz, almidón de maíz o harina de arroz, reaparecía el apetito en los niños y mejoraba la absorción de grasas haciendo desaparecer la diarrea grasa.**



- ....a partir de 1950 el tratamiento de pacientes celíacos se ha basado en la dieta libre de gluten

Celiac disease: pathogenesis of a model immunogenetic disease

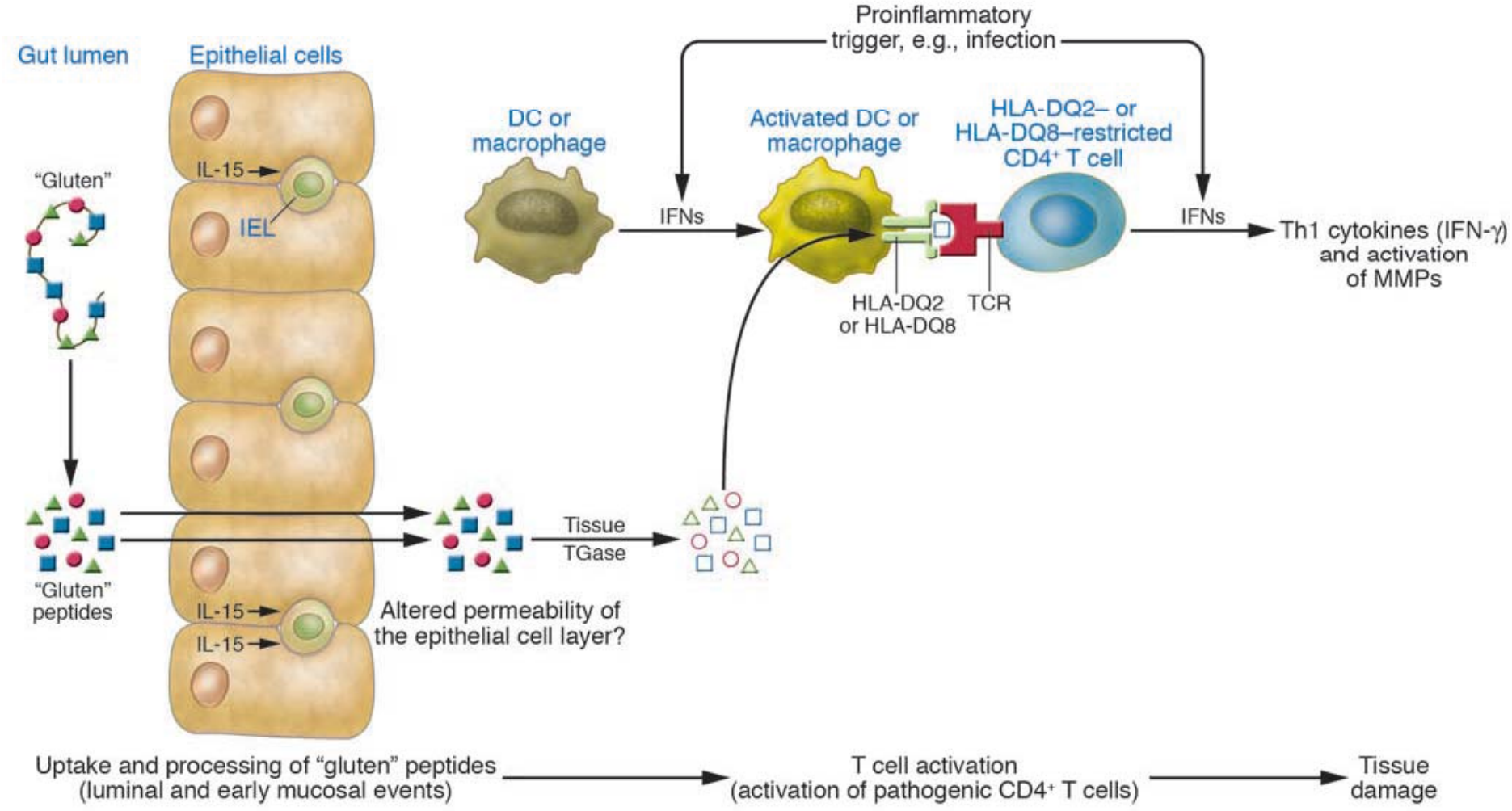
Martin F. Kagnoff

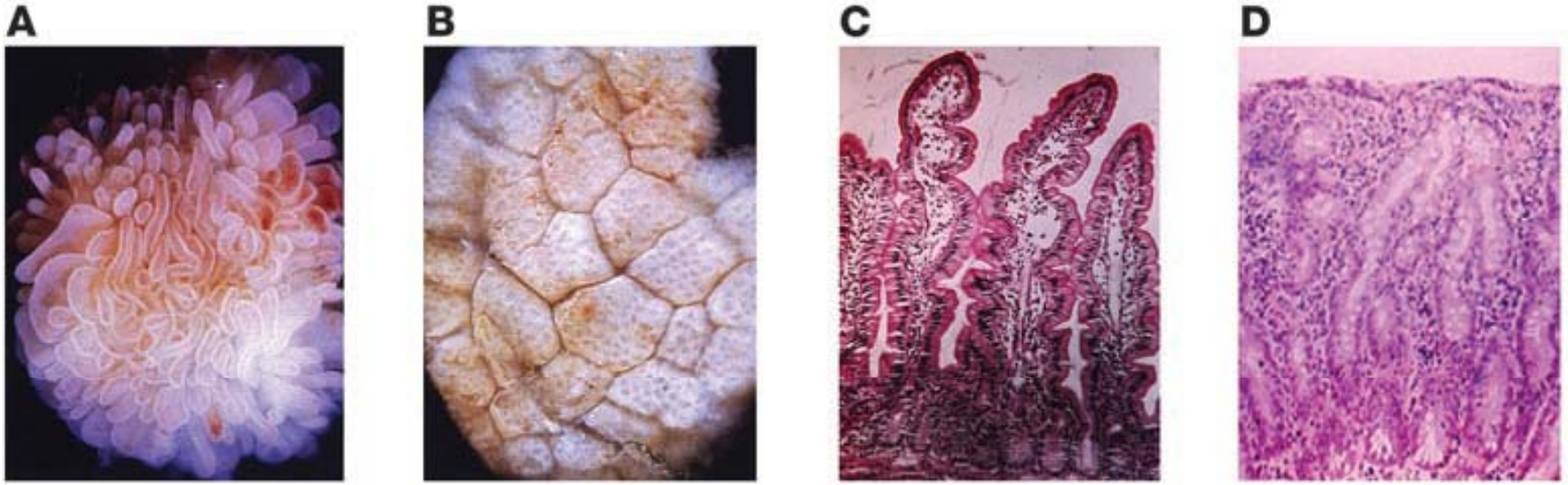
Departments of Medicine and Pediatrics, Laboratory of Mucosal Immunology, and Wm. K. Warren Medical Research Center for Celiac Disease, UCSD, La Jolla, California, USA.

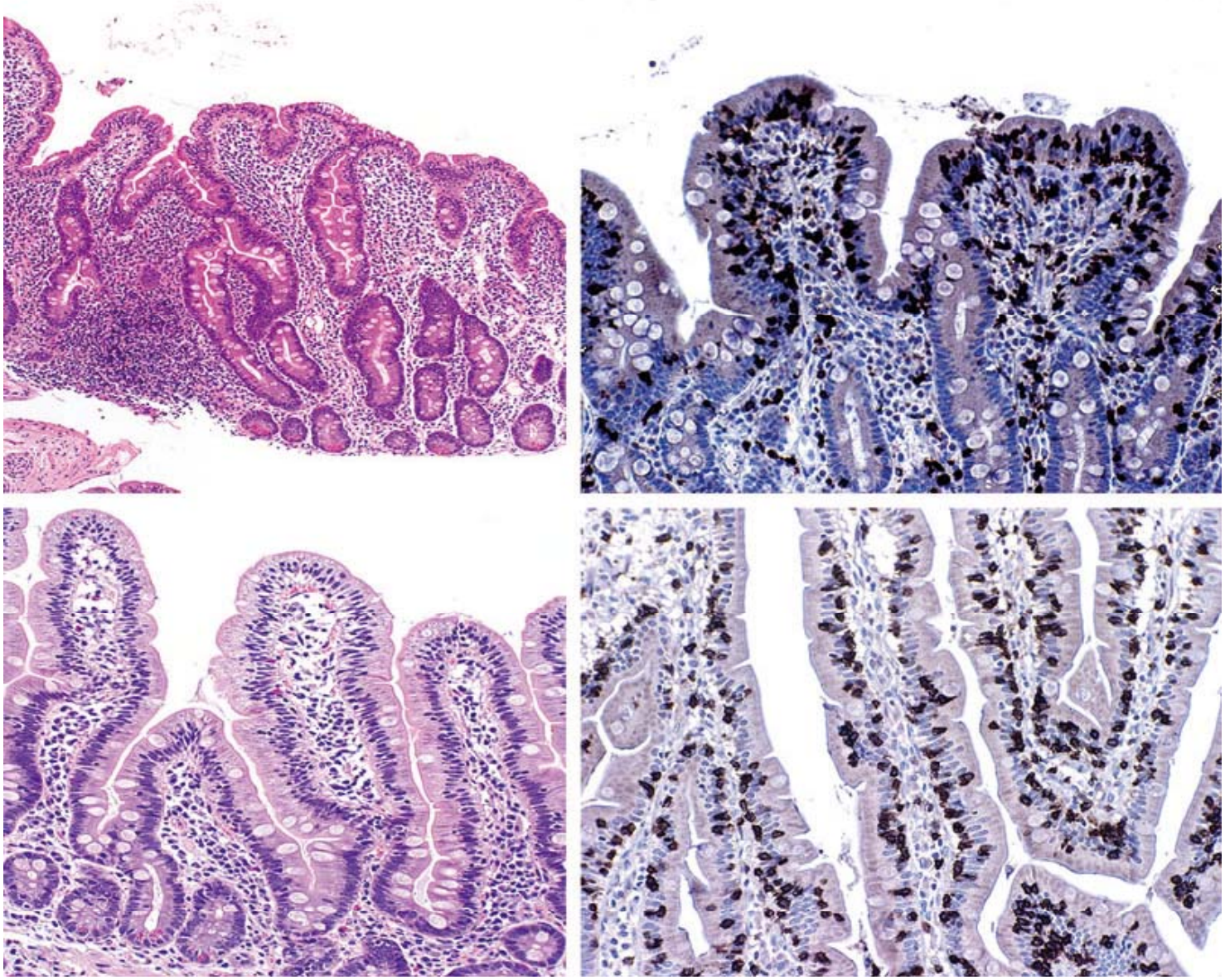
Celiac disease is characterized by small-intestinal mucosal injury and nutrient malabsorption in genetically susceptible individuals in response to the dietary ingestion of wheat gluten and similar proteins in barley and rye. Disease pathogenesis involves interactions among environmental, genetic, and immunological factors. Although celiac disease is predicted by screening studies to affect approximately 1% of the population of the United States and is seen both in children and in adults, 10%–15% or fewer of these individuals have been diagnosed and treated. This article focuses on the role of adaptive and innate immune mechanisms in the pathogenesis of celiac disease and how current concepts of immunopathogenesis might provide alternative approaches for treating celiac disease.

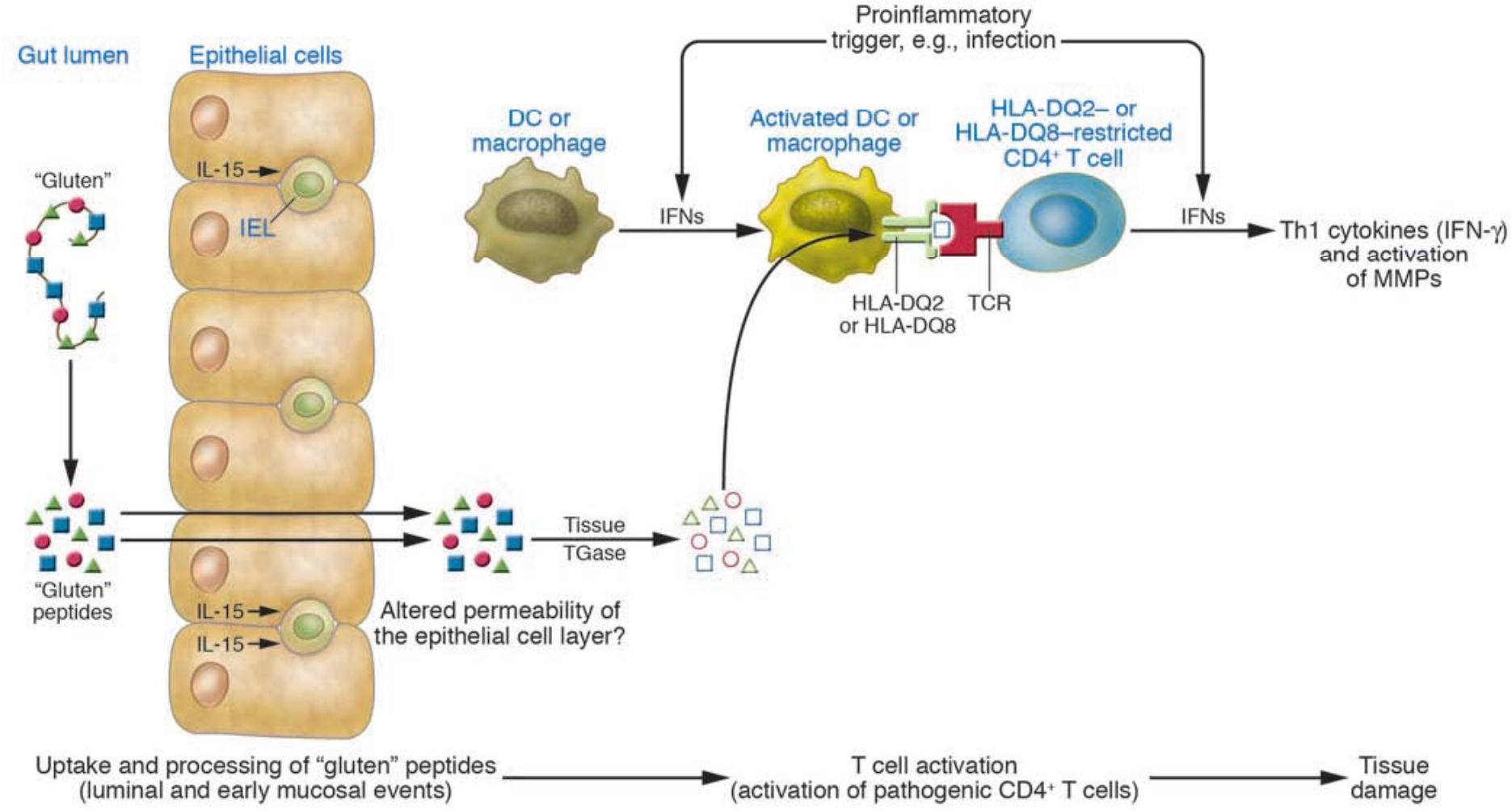
Celiac disease (CD) is characterized by small-intestinal mucosal injury and nutrient malabsorption. It is activated in genetically susceptible individuals by the dietary ingestion of proline- and glutamine-rich proteins that are found in wheat, rye, and barley and are widely termed "gluten" (1). Although approximately 1% of the population of the United States is affected by CD, most affected individuals remain undiagnosed. This probably reflects the fact that patients with CD can manifest a spectrum of intestinal

severe CD (Figure 1B) manifests a complete loss of villi, with a flat mucosal surface accentuated by ridges and numerous crypt openings. When tissue sections of the mucosa of the small intestine are stained with H&E, to visualize mucosal structure and the individual cells, the mucosa of healthy individuals is characterized by tall villi lined by a single layer of columnar epithelial cells with nuclei located near the basal surface; a smattering of intraepithelial lymphocytes (IELs) (approximately 1 per 6–10 epithelial cells);


Gut lumen
Epithelial cells
Proinflammatory
trigger, e.g., infection
DC or
macrophage
Activated DC or
macrophage
HLA-DQ2– or
HLA-DQ8–restricted
CD4+ T cell
"Gluten"
IL-15
IEL
IFNs
IFNs
Th1 cytokines (IFN-γ)
and activation
of MMPs
HLA-DQ2
or HLA-DQ8
TCR
"Gluten"
peptides
Tissue
TGase
IL-15
IL-15
Altered permeability of
the epithelial cell layer?
Uptake and processing of "gluten" peptides
(luminal and early mucosal events)
T cell activation
(activation of pathogenic CD4+ T cells)
Tissue
damage


A
B
C
D




Gut lumen
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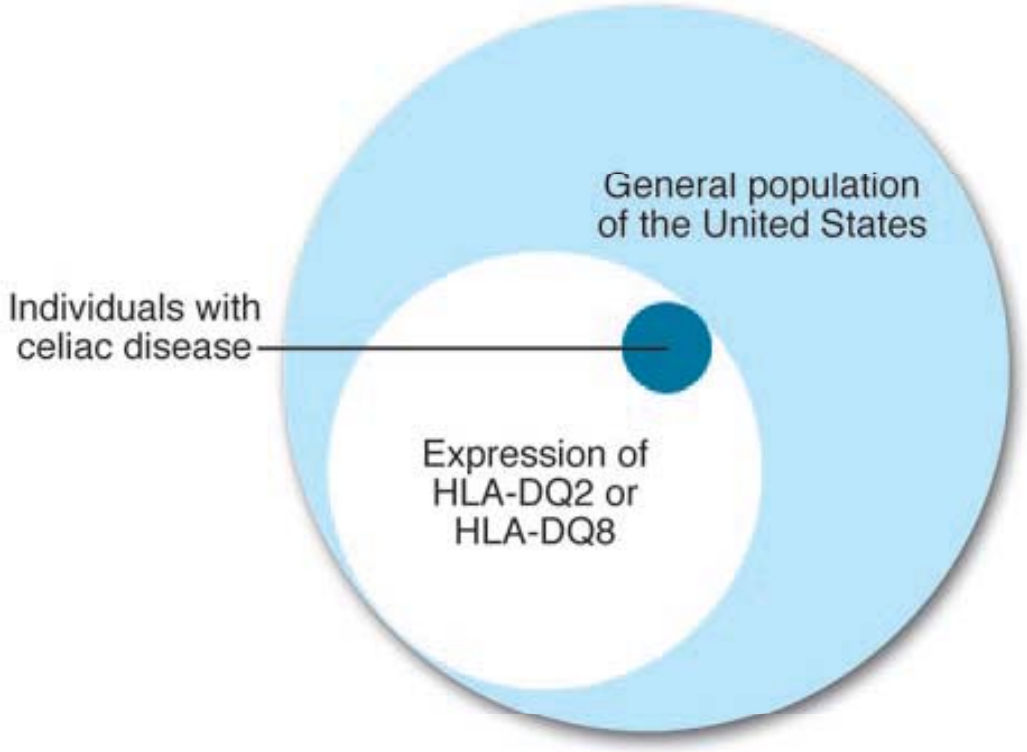
General population
of the United States
Individuals with
celiac disease
Expression of
HLA-DQ2 or
HLA-DQ8

World Journal of Gastroenterology

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wjg@wjgnet.com
doi:10.3748/wjg.v18.i37.5300

World J Gastroenterol 2012 October 7; 18(37): 5300-5304
ISSN 1007-9327 (print) ISSN 2219-2840 (online)

CASE REPORT

Origin of celiac disease: How old are predisposing haplotypes?

Giovanni Gasbarrini, Olga Rickards, Cristina Martínez-Labarga, Elsa Pacciani, Filiberto Chilleri, Lucrezia Laterza, Giuseppe Marangi, Franco Scaldaferri, Antonio Gasbarrini

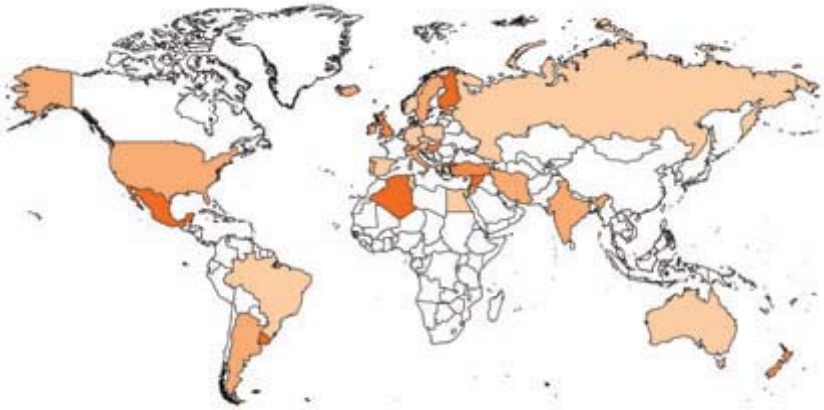
Giovanni Gasbarrini, Ricerca in Medicina Foundation NGO, Falcone and Borsellino Gallery, 40123 Bologna, Italy
Olga Rickards, Cristina Martínez-Labarga, Center of Molecular Anthropology for Ancient DNA Studies, Department of Biology, University of Rome Tor Vergata, 00173 Rome, Italy
Elsa Pacciani, Filiberto Chilleri, Superintendence for the Archaeological Heritage of Tuscany, 50018 Florence, Italy
Lucrezia Laterza, Franco Scaldaferri, Antonio Gasbarrini,

the first report showing the presence of a HLA haplotype compatible for CD in archaeological specimens.

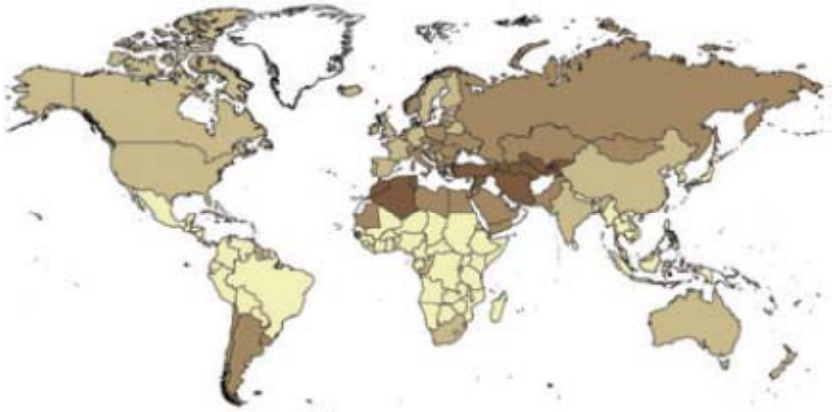
Key words: Celiac disease; Human leukocyte antigen haplotype; Ancient DNA; Single nucleotide polymorphisms;

Hallazgos arqueológicos.
Ciudad de Cosa- Sur de Italia..

Prevalence of celiac disease



Wheat consumption



Haplotype frequency of DR3-DQ2

Haplotype frequency of DR4-DQ8

Patrón clásico de la enfermedad

PANMALABSORCIÓN

GLÚCIDOS

Reducción depósitos de glucógeno hepático y muscular.

Aumento de producción de gas intestinal derivado de la fermentación de carbohidratos no absorbidos: diarrea explosiva

Sobreproducción de ácido láctico derivado de la fermentación de azúcares no absorbidos: diarrea osmótica.

PROTEÍNAS

Pérdida de peso y emaciación muscular.

Ascitis y edemas debidos a hipoalbuminemia.

Síntesis inadecuada de proteínas: defecto en la síntesis de las hormonas hipofisiarias: panhipopituitarismo.

GRASAS

Esteatorrea

Malabsorción de vitaminas liposolubles (A,D, K y E).

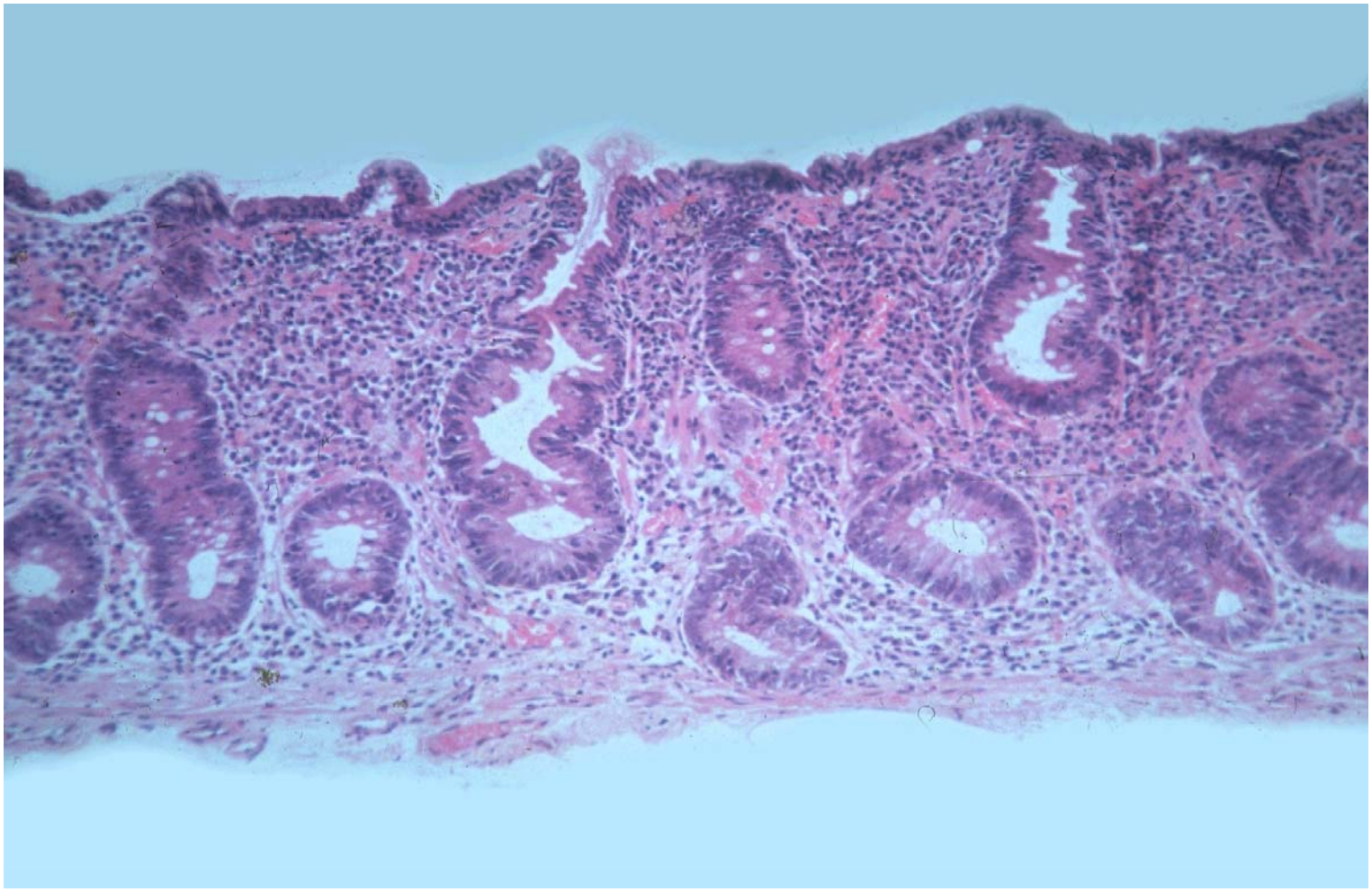
Síntomas dependientes de la malabsorción de vitaminas, minerales y oligoelementos.

Fe	Glositis	Potasio	Calambes musculares
	Coiloniquia		
	Anemia microcítica		Debilidad
Ácido fólico y Vit B12	Glositis	Zn	Acrodermatitis
	Estomatitis		
	Anemia macrocítica	Vitamina A	Hemeralopia
			Xerolftalmia
Calcio y vitamina D	Osteomalacia		Hiperqueratosis folicular
	Osteopenia		
	Osteoporosis		
	Dolores óseos	Vitamina K	Hipoprotrombinemia
	Fracturas espontáneas		Diatesis hemorrágica
		Vitamina B12	Neuropatía periférica
Calcio y magnesio		Tiamina	
	Tetania, parestesias		

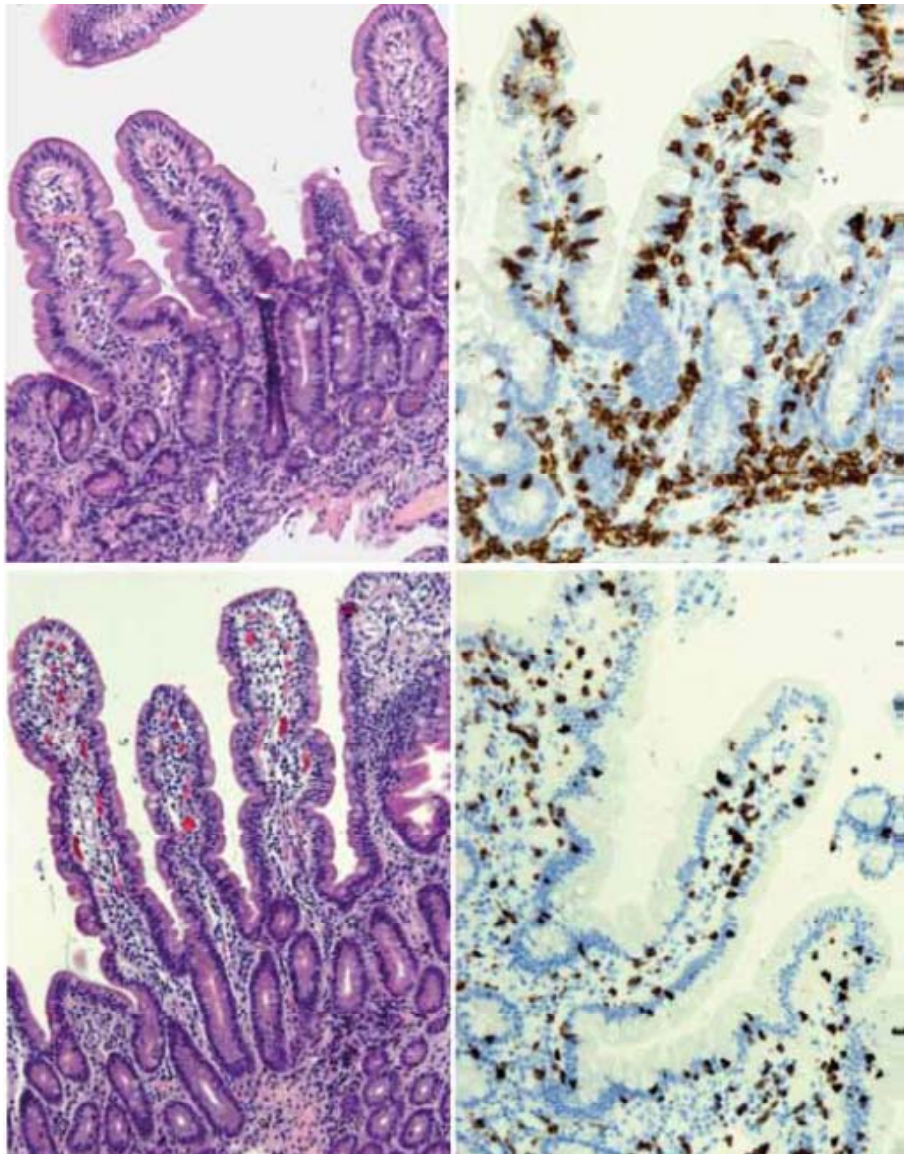
Emaciación


ZARAGOZA. Facultad de Medicina y Ciencias

Atrofia grave de las vellosidades



Varón de 21 años que desde hace varios meses presenta sensación de saciedad precoz, digestiones pesadas e hinchazón, después de las comidas.




I'm Jennifer Esposito.
I have celiac disease.

PEDIATRICS
OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

The Changing Face of Childhood Celiac Disease in North America: Impact of Serological Testing

Kelly E. McGowan, Derek A. Castiglione and J. Decker Butzner

Pediatrics 2009;124;1572-1578
DOI: 10.1542/peds.2008-2373

American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN™

PRESENTATION	Pretesting (n: 36)	Testing (N: 199)	*p*
CLASSIC	24 (67)	30 (19)	< 0.01
ATYPICAL GASTROINTESTINAL	7 (19)	75 (38)	0.482
Abdominal pain plus other	5	34	
Abdominal pain only	0	18	
Endoscopy for other reason	0	8	
Chronic diarrhea	1	7	
Constipation	0	5	
Vomiting	1	2	
Food allergy	0	1	
Abdominal distension	0	1	
EXTRAINTESTINAL	5 (14)	29 (15)	0.914
Failure to thrive	2	13	
Iron deficiency with o without anemia	2	6	
Short stature	0	6	
Dermatitis herpetiformis	0	2	
Elevated transaminase levels	0	1	
Dental enamel defects	0	1	
Hypoalbuminemia	1	0	
SILENT	0(0)	55 (28)	< 0.01
Familiy history	0	35	
Tipe 1 diabetes mellitus	0	14	
Trisomy 21	0	5	
Hypothiroidism	0	1	

European Society for Pediatric Gastroenterology, Hepatology, and Nutrition Guidelines for the Diagnosis of Coeliac Disease

*S. Husby, †S. Koletzko, ‡I.R. Korponay-Szabó, §M.L. Mearin, ||A. Phillips, ¶R. Shamir, #R. Troncone, **K. Giersiepen, ††D. Branski, ‡‡C. Catassi, §§M. Lelgeman, ||||M. Mäki, ¶¶C. Ribes-Koninckx, ##A. Ventura, and ****K.P. Zimmer, for the ESPGHAN Working Group on Coeliac Disease Diagnosis, on behalf of the ESPGHAN Gastroenterology Committee

ABSTRACT

Objective: Diagnostic criteria for coeliac disease (CD) from the European Society for Paediatric Gastroenterology, Hepatology, and Nutrition (ESPGHAN) were published in 1990. Since then, the autoantigen in CD, tissue transglutaminase, has been identified; the perception of CD has changed from that of a rather uncommon enteropathy to a common multiorgan disease strongly dependent on the haplotypes human leukocyte antigen (HLA)-DQ2 and HLA-DQ8; and CD-specific antibody tests have improved.
Methods: A panel of 17 experts defined CD and developed new diagnostic criteria based on the Delphi process. Two groups of patients were defined with different diagnostic approaches to diagnose CD: children with symptoms suggestive of CD (group 1) and asymptomatic children at increased risk for CD (group 2). The 2004 National Institutes of Health/

Results: In group 1, the diagnosis of CD is based on symptoms, positive serology, and histology that is consistent with CD. If immunoglobulin A anti-tissue transglutaminase type 2 antibody titers are high (>10 times the upper limit of normal), then the option is to diagnose CD without duodenal biopsies by applying a strict protocol with further laboratory tests. In group 2, the diagnosis of CD is based on positive serology and histology. HLA-DQ2 and HLA-DQ8 testing is valuable because CD is unlikely if both haplotypes are negative.
Conclusions: The aim of the new guidelines was to achieve a high diagnostic accuracy and to reduce the burden for patients and their families. The performance of these guidelines in clinical practice should be evaluated prospectively.

Down Syndrome and Celiac Disease.

- AB Summary: Down syndrome is associated with immune-related disorders such as hypothyroidism, insulin-dependent diabetes mellitus, and celiac disease. In this study we determined antigliadin antibodies (AGA) in 54 patients with Down syndrome; 22 had AGA values above the cutoff limit. Nineteen patients underwent intestinal biopsy, and total or subtotal villous atrophy was found in nine. There was a total of 65 patients with Down syndrome in our area of southern Sweden; two were already known to have celiac disease. **The minimum prevalence of celiac disease in Down syndrome in this area in southern Sweden was 11 of 65 or 16.9%.** (C) Lippincott-Raven Publishers.

Prevalence of Celiac Disease in Down Syndrome in the United States.

- AB Background: Numerous studies in Europe have documented a high prevalence of celiac disease in Down syndrome. This study was undertaken to estimate the prevalence of celiac disease in Down syndrome in the southeastern United States. Methods: Seventy-five patients with Down syndrome were screened using immunoglobulin (Ig)A-anti antiendomysium antibodies, IgA-antigliadin antibodies, and total IgA level. When either antiendomysium or antigliadin antibodies produced positive findings, patients were referred to a pediatric gastroenterologist for consideration of a duodenal biopsy. Results: Thirteen percent (10/75) were positive for antiendomysium antibodies. Half of these patients were also positive for antigliadin antibodies. Six of 10 patients positive for antiendomysium antibodies underwent intestinal biopsy. Changes consistent with celiac disease were documented in five. Histologic findings ranged from focal to total villous atrophy. None had IgA deficiency. Conclusions: There was a high prevalence of positivity to antiendomysium antibody in Down syndrome. Antiendomysium antibody was a more sensitive screening test than antigliadin antibody. **The prevalence of celiac disease in Down syndrome in the southeastern United States was 1 in 14 cases**. Screening with antiendomysium antibody and IgA for all children with Down syndrome is recommended, even if there are no gastrointestinal symptoms. (C) 2000 Lippincott Williams & Wilkins, Inc.

Prevalence and Clinical Picture of Celiac Disease in Italian Down Syndrome Patients: A Multicenter Study.

- AB Background: A multicenter research study of Down syndrome patients was carried out to estimate the prevalence of celiac disease in patients with Down syndrome and to show clinical characteristics and laboratory data of Down syndrome patients. Methods: The authors studied 1,202 Down syndrome patients. Fifty-five celiac disease patients (group 1) were compared with 55 immunoglobulin A antigliadin-positive antiendomysium antibodies-negative patients (group 2) and with 57 immunoglobulin A antigliadin-negative antiendomysium antibodies-negative patients (group 3). Results: Celiac disease was diagnosed in 55 of 1,202 Down syndrome patients (4.6%). In group 1, weight and height percentiles were shifted to the left, whereas these parameters were normally distributed in groups 2 and 3. In celiac patients, diarrhea, vomiting, failure to thrive, anorexia, constipation, and abdominal distension were higher than in the other two groups. Low levels of hemoglobinemia, serum iron, and calcium were observed more frequently in group 1**. The diagnosis of celiac disease was made after a mean period of 3.8 years from the initiation of symptoms. Sixty-nine percent of patients showed a classic presentation, 11% had atypical symptoms, and 20% had silent celiac disease.** Autoimmune disorders were more frequent (30.9%) in group 1 than in the other two groups examined (15%;$P < 0.05$). Conclusions: This study reconfirms a high prevalence of celiac disease in Down syndrome. However, the diagnostic delay, the detection of atypical symptoms or silent form in one third of the cases, and the increased incidence of autoimmune disorders suggest the need for the screening of celiac disease in all Down syndrome patients. (C) 2001 Lippincott Williams & Wilkins, Inc.

Tissue Transglutaminase Antibodies Are a Useful Serological Marker for the Diagnosis of Celiac Disease in Patients With Down Syndrome.

- AB Background: Celiac disease (CD) is overrepresented among patients with Down syndrome (DS), who frequently lack any typical symptoms. Therefore, screening for CD is recommended in this high-risk group. The aim of the study was to determine the prevalence of CD in Arab children with DS and evaluate the contribution of immunoglobulin (Ig) A and IgG anti-gliadin antibodies (AGA), IgA and IgG tissue transglutaminase (TTG) antibodies, and IgA anti-endomysial antibodies (EMA) to screen for CD in children with DS. Patients and Methods: A total of 52 Arab patients with DS and 52 healthy Arab control subjects were studied for CD using various serological markers. Data on age, sex, weight, height, gastrointestinal symptoms, and endocrine abnormalities were recorded. Human leukocyte antigen (HLA) was studied in patients undergoing small intestinal biopsy. Results: Five patients with DS were IgA TTG-positive and only 1 patient with DS was IgG TTG-positive. EMA was negative in all patients with DS. TTG (IgA and IgG) and EMA were negative in all control children. IgA AGA was positive in 12 patients with DS and 3 control subjects (P = 0.02), whereas IgG AGA was positive in 41 patients with DS and 26 control subjects (P = 0.004). Only children testing positive for TTG underwent upper endoscopy with duodenal biopsy. Two children with DS were diagnosed with CD. Both patients were IgA TTG-positive. One was HLA DQ2-positive and another was negative for HLA DQ2 and DQ8. Conclusions: **CD is prevalent (3.8%) in Arab patients with DS**. Based on our cohort, IgA TTG is useful in diagnosing patients with CD and DS. (C) 2007 Lippincott Williams & Wilkins, Inc.

Gut 1997; **40:** 492–496

Down's syndrome is strongly associated with coeliac disease

L Gale, H Wimalaratna, A Brotodiharjo, J M Duggan

Abstract

***Background*—There is evidence of an increased prevalence of coeliac disease in Down's syndrome.**

***Aims*—To investigate the association, patients with Down's syndrome and matched controls were examined.**

***Methods*—Fifty nine patients with Down's syndrome residing in government institutions in the Hunter region of New South Wales were studied. Four were excluded**

43 times that of other children; another[10] suggested a 20-fold increase.

The next progression has been to prospectively survey populations of patients with Down's syndrome, using antibody screening tests to determine eligibility for biopsy.[11–16] It is difficult to draw conclusions about the strength of the association between coeliac disease and Down's syndrome from these studies because of the lack of comparability in the findings (Table I). The patients have

See 1 citation found using an alternative search:

Eur J Gastroenterol Hepatol. 2001 Mar;13(3):263-7.

Prevalence of coeliac disease in Down's syndrome.

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Abstract

OBJECTIVE: During the past decade, it has been shown that the association between Down's syndrome and coeliac disease is relatively frequent Prevalence rates of coeliac disease in patients with Down's syndrome reported by different authors are significantly higher than those found in the general population. The main purpose of this study was to assess the prevalence of coeliac disease in a series of subjects with Down's syndrome from our geographical area.

DESIGN: A cross-sectional study.

SETTING: Outpatient paediatric clinics of acute-care teaching hospitals in Barcelona, Spain.

PARTICIPANTS: A total of 284 persons with Down's syndrome aged between 1 and 25 years were included in the study. In all cases, serum concentrations of antigliadin antibodies (AGAs) (Pharmacia CAP system enzyme-linked immunosorbent assay), antiendomysium antibodies (AEA) (indirect immunofluorescence) of immunoglobulin (Ig)A class or IgG class in cases of IgA deficiency were determined. Jejunal biopsy was offered to all patients with AEA positivity and to those with suggestive clinical manifestations of coeliac disease. In all patients, a clinical study was made to evaluate the presence and time-course of symptoms related to coeliac disease.

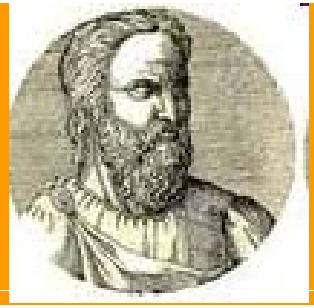
MAIN OUTCOME MEASURES AND RESULTS: In 18 of the 284 subjects with Down's syndrome, aged between 2 and 15 years, coeliac disease was confirmed by jejunal biopsy. Accordingly, the minimum prevalence rate of coeliac disease was of 6.3%. Ninety-four percent (17/18) and 78% (14/18) of patients with the association Down's syndrome and coeliac disease showed AEA and AGA positivity, respectively. Fifteen patients with the association coeliac disease and Down's syndrome (15/18) showed clinical manifestations compatible with coeliac disease, with a predominance of intestinal symptoms (8/18) over those with atypical or extra-intestinal forms (7/18). Three patients had clinically silent forms of coeliac disease (3/ 18).

CONCLUSIONS: Measurement of serum concentrations of AEA should be added to the list of screening tests for coeliac disease in patients with Down's syndrome, otherwise definite association between both diseases may pass unnoticed and diagnosis of coeliac disease be considerably delayed.

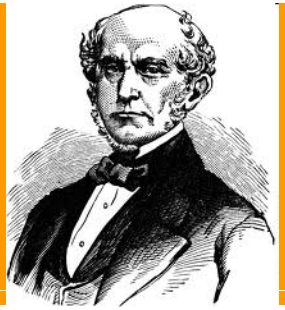
PMID: 11293446 [PubMed - indexed for MEDLINE]

⊕ **Publication Types, MeSH Terms**

⊕ **LinkOut - more resources**



BASE « ARETÆUS »



« ARETÆUS »

Módulos del Registro (I)

1 **TRAYECTORIA DEL PACIENTE ANTES DEL DIAGNÓSTICO**

- Tiempo de evolución — **1a**
- Consumo de recursos — **1b**

2 **PATRONES DE PRESENTACIÓN**

3 **TRAYECTORIA DEL ENFERMO CON BUENA RESPUESTA AL TRATAMIENTOº**

4 **RESULTADO DE LOS PACIENTES CON RESPUESTA PARCIAL**

5 **INVESTIGACIÓN DE LOS NO RESPONDEDORES**

6 **CELÍACOS REFRACTARIOS**

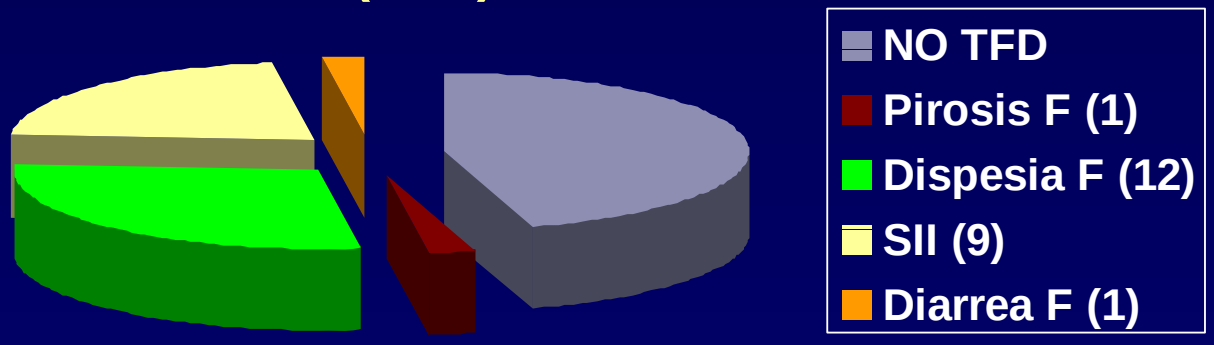
Resultados: Aspectos Clínicos

Tiempo de Evolución de los Síntomas:

- Mediana: 120 meses (rango: 2-600 meses)
- <18 meses: 8 casos (20,5%); 18-60 meses: 10 casos (25,5%); >60 meses: 21 casos (54%)]

Antecedente de diagnóstico de TFD:

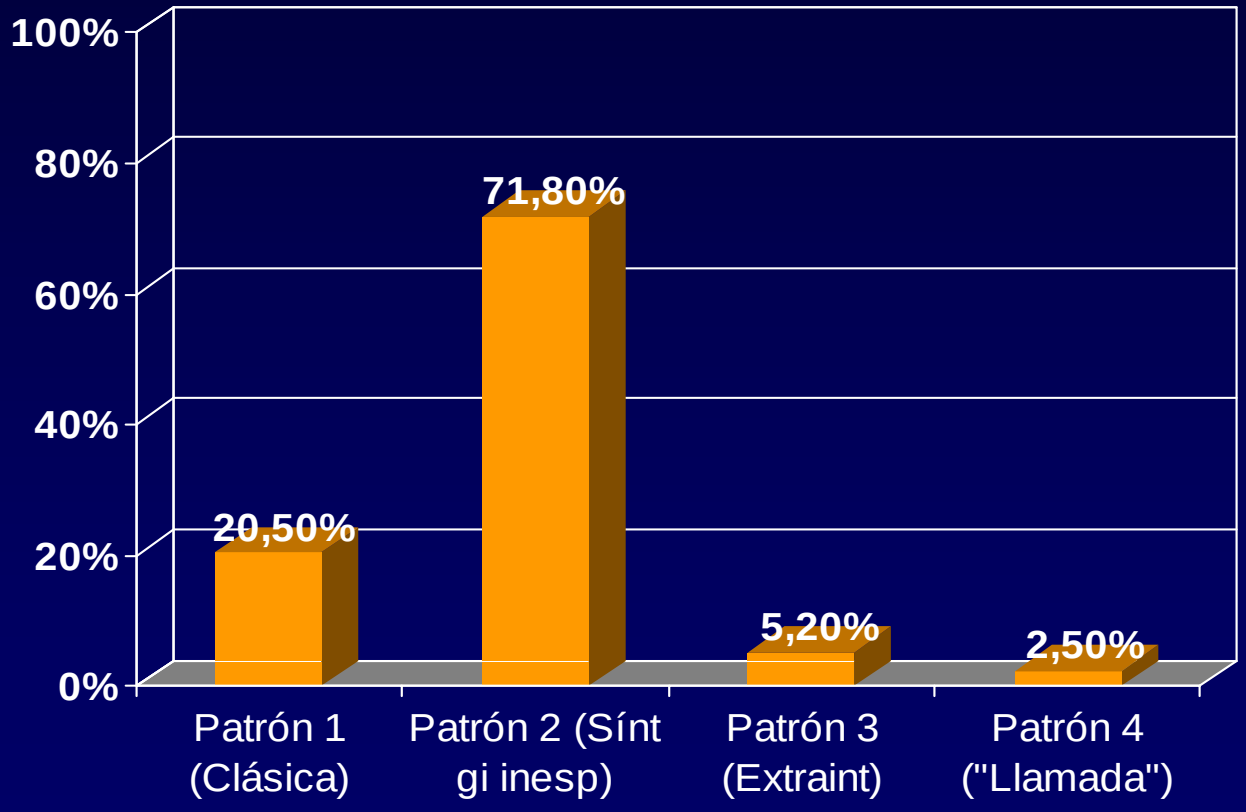
Un total de 23 casos **(59%).**



El 100% de los casos presentaron síntomas g.i inespecíficos.

Resultados: Aspectos Clínicos

Patrones de Presentación Clínica (motivo de consulta):



Patrón 1	8 casos
Patrón 2	28 casos
Patrón 3	2 casos
Patrón 4	1 caso

36 casos (92%) presentaron manifest. extraintest. relacionadas con ESG

Resultados: Aspectos Clínicos

Antecedentes Familiares de ESG o de enfermedades relacionadas: Implicaciones en la demora diagnóstica

Promedio de tiempo en meses desde el inicio de los síntomas hasta el diagnóstico, en función de la existencia de AF de enfermedades relacionadas con la ESG. (“n” de cada grupo)

	AF de al menos una enfermedad relacionado con ESG	AF de ESG	AF de EII	AF de enfermedad Autoinmune
SI	106,62 (n=16)	97,08 (n=13)	**210 (n=2)**	126,5 (n=6)
NO	**181,04 (n=23)**	**177,23 (n=26)**	147,29 (n=37)	**154,88 (n=33)**

p>0,05

Resultados: Aspectos Económicos

Consumo de recursos previo al diagnóstico de ESG

- Visitas médicas: 38 casos (97,5%)
- Fármacos:

IBP: 21 (54%)	Procinéticos: 8 (20,5%)	Antiácidos: 9 (23%)
Laxantes: 6 (15,4%)	Antidiarreicos: 4 (10,2%)	Espasmolíticos: 2 (5,1%)
Sales ferrosas: 6 (15,4%)		Psicotropos: 2 (5,1%)

- Pruebas de laboratorio: 35 casos (90%)
- Pruebas Rx: 19 pacientes (48,7%). En 18, pruebas básicas (RX simple, Eco, estudios baritados). En 3 casos, TC o RM.
- Endoscopia: 21 casos (54%). Alta: 15 (38,5%). Baja: 11 (28%).

Resultados: Aspectos Económicos

Costes imputables a pruebas complementarias

- Coste medio por caso: **210,31** euros. (DE=207,74)

Costes promedio en concepto de exámenes complementarios, distintos de los que permitieron establecer el diagnóstico de ESG (en euros), y "n" de cada grupo.

Tº evolución de los síntomas	Patrón presentación clínica	Diagnóstico previo de TFD	AF relacionados con la ESG
< 18 meses: 88,9 (n= 8)	**Patrón 1: 308,8 (n=7)**	TFD: 227,6 (n=23)	ESG: 157,9 (n=13)
18-60 meses: 294,5 (n=10)	Patrón 2: 156,6 (n=28)	**TFD alto: 254,84 (n=13)**	EII: 223 (n=2)
>60 meses: 216,5 (n=21)	Patrón 3: 0 (n=2)	TFD bajo: 192,2 (n=10)	Enf. autoinmune: 157,7 (n=6)
	Patrón 4: 0 (n=1)	Ningún TFD: 185,4 (n=16)	**Ningún AF: 237,6 (n=23)**

p>0,05

¿ Cómo sospechar la enfermedad?

- Síntomas gastrointestinales inespecíficos
  - Sensación de saciedad precoz, plenitud después de las comidas, digestiones lentas y pesadas..
  - Flatulencia, distensión abdominal y meteorismo.
  - Cambios frecuentes en el ritmo intestinal (diarrea, estreñimiento)
  - Crisis episódicas de dolor intenso en el estómago con náuseas y vómitos.
- Síntomas extraintestinales:
  - Retraso en el crecimiento y desarrollo
  - Fracturas ante traumatismos mínimos.
  - Aftas orales.
  - Lesiones en la piel tipo psoriasis, ezcemas, acné.
  - Anemia o ferropenia de origen no aclarado.
  - Cambios frecuentes en el estado de ánimo, irritabilidad, distimia, depresión.
  - Elevación inexplicable de transaminasas.
  - Epilepsia