

ThermoFisher SCIENTIFIC

Recognising coeliac disease earlier in primary care

Agenda

1. Introduction
2. What you need to know about coeliac disease
3. Who should be tested?
4. Guidelines for diagnosis and management of coeliac disease
5. Summary





Introduction

Introduction to coeliac disease through patient cases

Patient case 1: Chronic gastrointestinal symptoms with weight loss[1]

History:

Female, 37 years old, with a 10-year history:

- Irritable bowel disease & dyspepsia
- Recurrent diarrhoea and bloating

Now: abdominal distension and recent weight loss



Laboratory results*:

* Laboratory results converted to International System on Units (SI) where appropriate.

Normal:

C-reactive protein, thyroid and liver function tests, glomerular filtration rate and albumin

Abnormal – malabsorption pattern:

- Haemoglobin 100 g/L (anaemia)
- Mean corpuscular volume 69 fL (microcytic)
- Ferritin 1 µg/L
- Vitamin B12 59 pmol/L
- Vitamin D 20 nmol/L
- Anti-tTG IgA antibodies 63 U/mL (ref 0–12)

Gastroenterology:

Endoscopy:

Scalloped duodenal folds

Biopsy:

Villous atrophy
Crypt hyperplasia
Lymphocytic infiltration

Diagnosis:

Coeliac disease

1. Atak BT, et al. A case of celiac disease admitted to our clinic with symptoms of malabsorption. Natl J Health Sci. 2023;8(1):40-41.

Introduction to coeliac disease through patient cases

Patient case 2: Severe malabsorption with behavioral changes[1]

History:

Male, 25 years old

Autism spectrum disorder + schizophrenia
Since 1–2 months: vomiting and diarrhoea
14–18 kg weight loss (12 months)

Behavioural changes:

- Agression
- Severe disturbances
- Eating constantly



Laboratory results*

- Potassium 2.8 mmol/L
- Magnesium 0.45 mmol/L
- Albumin 18 g/L
- Folate 6.8 nmol/L
- Vitamin D <10 nmol/L
- Vitamin A 0.98 µmol/L
- Vitamin E 5.3 µmol/L
- Zinc 3.5 µmol/L
- Copper 9.9 µmol/L
- Anti tTG IgA antibodies >250 U/mL

* Laboratory results converted to International System on Units (SI) where appropriate.

Gastroenterology

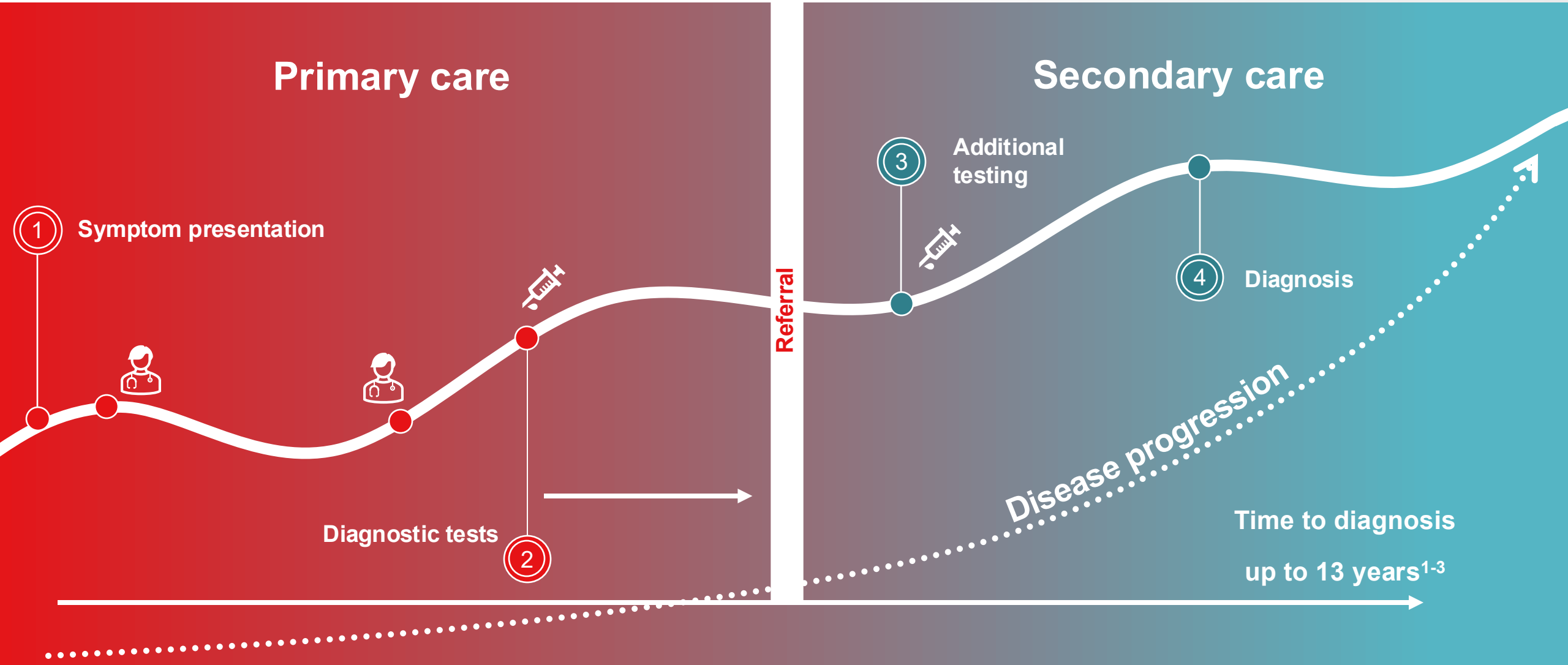
- **Duodenal biopsy:**
  Marked villous blunting
  Marsh type 3b

Diagnosis:

Coeliac disease

1. Murphy AK, et al. Celiac disease in an adult presenting as behavioral disturbances: rare coexistence of disease or pathology. Am J Case Rep. 2020;21:e928337

The long road to coeliac disease diagnosis for a patient



1. Kårhus LL, et al. Diagnostic delay in coeliac disease: a survey among Danish patients. Can J Gastroenterol Hepatol. 2022;2022:5997624.
2. Violato M, et al. The impact of diagnosis on health-related quality of life in people with coeliac disease: a UK population-based longitudinal perspective. BMC Gastroenterol. 2019;19(1):68.
3. Fuchs V, et al. Delayed celiac disease diagnosis predisposes to reduced quality of life and incremental use of health care services and medicines: A prospective nationwide study. United European Gastroenterology Journal. 2018;6(4):567-575.

What is the challenge for a general practitioner?

Up to 12% of patients presenting at primary care have a functional gastrointestinal disorder (FGID)[1]

FGID has a worldwide prevalence of 40%[1]



Recurrent gastrointestinal (GI) distress[1-4]

- Coeliac disease
- Infectious diseases
- Colon cancer
- Irritable bowel syndrome
- Inflammatory bowel disease
- Lactose/fructose intolerance
- Food allergy

* IBS belongs to the group of functional bowel disorders (FBD)

If symptoms are unexplained or persistent despite usual care → consider testing for coeliac disease[5]

1. Fikree A, et al. Management of functional gastrointestinal disorders. Clin Med (Lond). 2021;21(1):44-52. 2. Rej A, et al. An update on coeliac disease from the NHS England National Centre for Refractory Coeliac Disease. Clinical Medicine (London, England). 2021;21(2):127-130 3. Caio G, et al. Celiac disease: a comprehensive current review. BMC Med. 2019;17(142):1-20. 4. Aziz I, et al. The overlap between irritable bowel syndrome and organic gastrointestinal diseases. Lancet Gastroenterol Hepatol. 2020;5(11):1095-1105. 5. National Institute for Health and Care Excellence (NICE). Coeliac disease: recognition, assessment and management (NG20). London: NICE; 2015



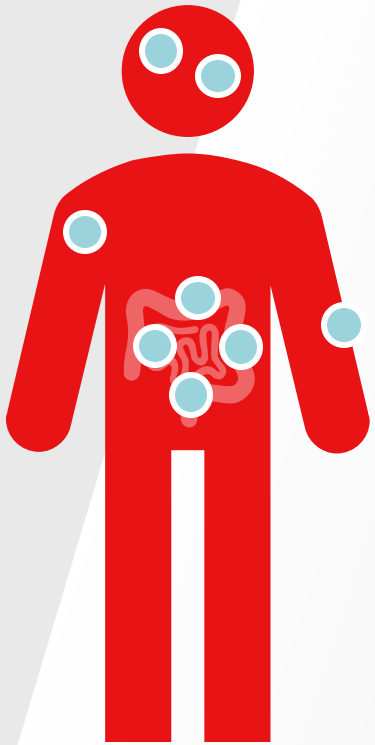
What you need to know about coeliac disease

What is coeliac disease?

Coeliac disease

A common **systemic autoimmune disease** caused by the ingestion of gluten, that can develop at any age[1,4]

- European seroprevalence is around 1%, with nearly 75% of cases remaining undiagnosed, according to a study of the Norwegian population[2,3]
- Incidence is rising[1,4]
- More prevalent in women[1,3]
- Presentation has changed over time[3]
- Risk factors include other autoimmune diseases, relatives with coeliac disease (CeD) and irritable bowel syndrome (IBS)[1–4]



Symptoms[1,3,4]

Head / mouth
- Anxiety
- Depression
- Mouth ulcers
- Enamel problems

Stomach
- Abdominal pain
- Vomiting
- Nausea

Intestinal
- Diarrhoea
- Constipation
- Flatulence

In adults
- Infertility
- Miscarriage
- Bone fractures

In children
- Failure to grow
- Short stature
- Delayed puberty
- Bone fractures

Other
- Anaemia
- Dermatitis herpetiformis

Coeliac disease often presents with extra-intestinal manifestations[3,4]

1. Al-Toma A, et al. European Society for the Study of Coeliac Disease 2025 Updated guidelines on the diagnosis and management of coeliac disease in adults. United European Gastroenterol J. 2025;00:1–32.
2. Kvamme JM, et al. Population-based screening for celiac disease reveals that the majority of patients are undiagnosed and improve on a gluten-free diet. Sci Rep. 2022;12(1):12647.
3. Lindfors K, et al. Coeliac disease. Nat Rev Dis Primers. 2019;5:3.
4. Rej A, et al. An update on coeliac disease from the NHS England National Centre for Refractory Coeliac Disease. Clin Med (Lond). 2021;21(2):127-130.

With 1% prevalence, what can you expect in your practice?[1,2]

How many patients might from in your primary care network might you see?

How many of these coeliac patients are diagnosed?

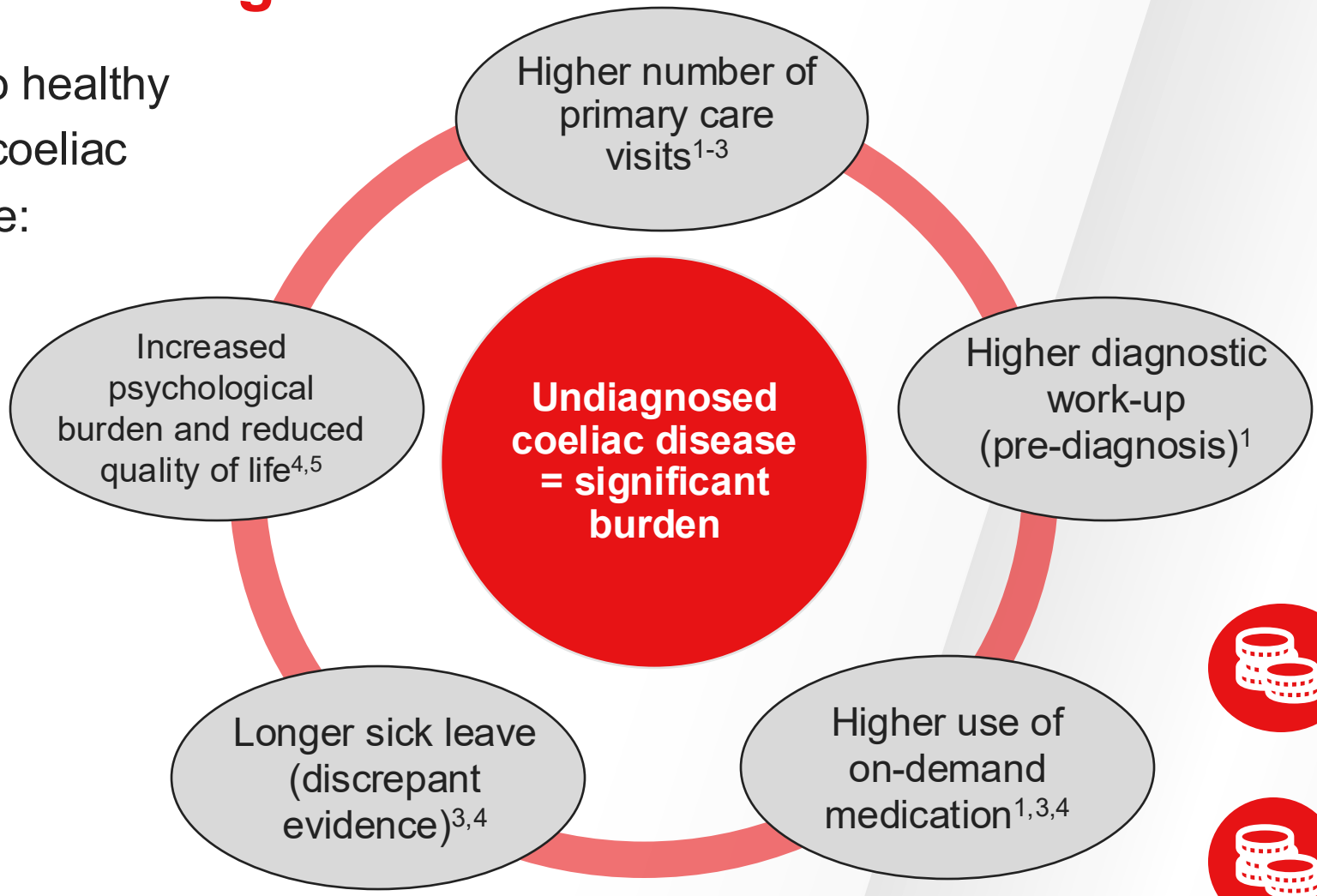
Expected coeliac patients = 21*

* Based on the mean number of patients in a practice

1. Singh P, et al. Global prevalence of celiac disease: systematic review and meta-analysis. *Clin Gastroenterol Hepatol.* 2018;16(6):823–836.
2. Eurostat. Healthcare personnel statistics – physicians [Internet]. Luxembourg: European Commission; [cited 2026 Mar 12]. Available from: https://ec.europa.eu/eurostat/statistics-explained/index.php?title=Healthcare_personnel_statistics_-_physicians

Burden of undiagnosed coeliac disease

Compared to healthy individuals, coeliac patients have:



Early diagnosis: possible healthcare cost savings over time[1,2]

Additional patient economical burden: food and social costs[4]

1. Violato M, et al. Resource use and costs associated with coeliac disease before and after diagnosis in 3,646 cases: results of a UK primary care database analysis. PLoS One. 2012;7(7):e41308.
2. Rahmanian M, et al. Estimating the costs of delayed Celiac disease diagnosis. BMC Gastroenterol. 2025;25:673.
3. Ukkola A, et al. Use of health care services and pharmaceutical agents in coeliac disease: a prospective nationwide study. Aliment Pharmacol Ther. 2020;51(6):1339-1345.
4. Bokemeyer B, et al. Burden of coeliac disease in Germany: real-world insights from a large retrospective health insurance claims database analysis. Therapeutic Advances in Gastroenterology. 2025;18.
5. Fjorback SSO, et al. 'It was hell on earth': perspectives of people living with celiac disease on diagnostic delay. Journal of Human Nutrition and Dietetics. 2024;37(6):1486-1495



Who should be tested?

Who should be tested: identifying high risk groups

For every diagnosed coeliac disease patient, three remain undiagnosed (75%).

So, who should you test?



1. Kvamme JM, et al. Population-based screening for coeliac disease reveals that the majority of patients are undiagnosed and improve on a gluten-free diet. Sci Rep. 2022;12:12647.

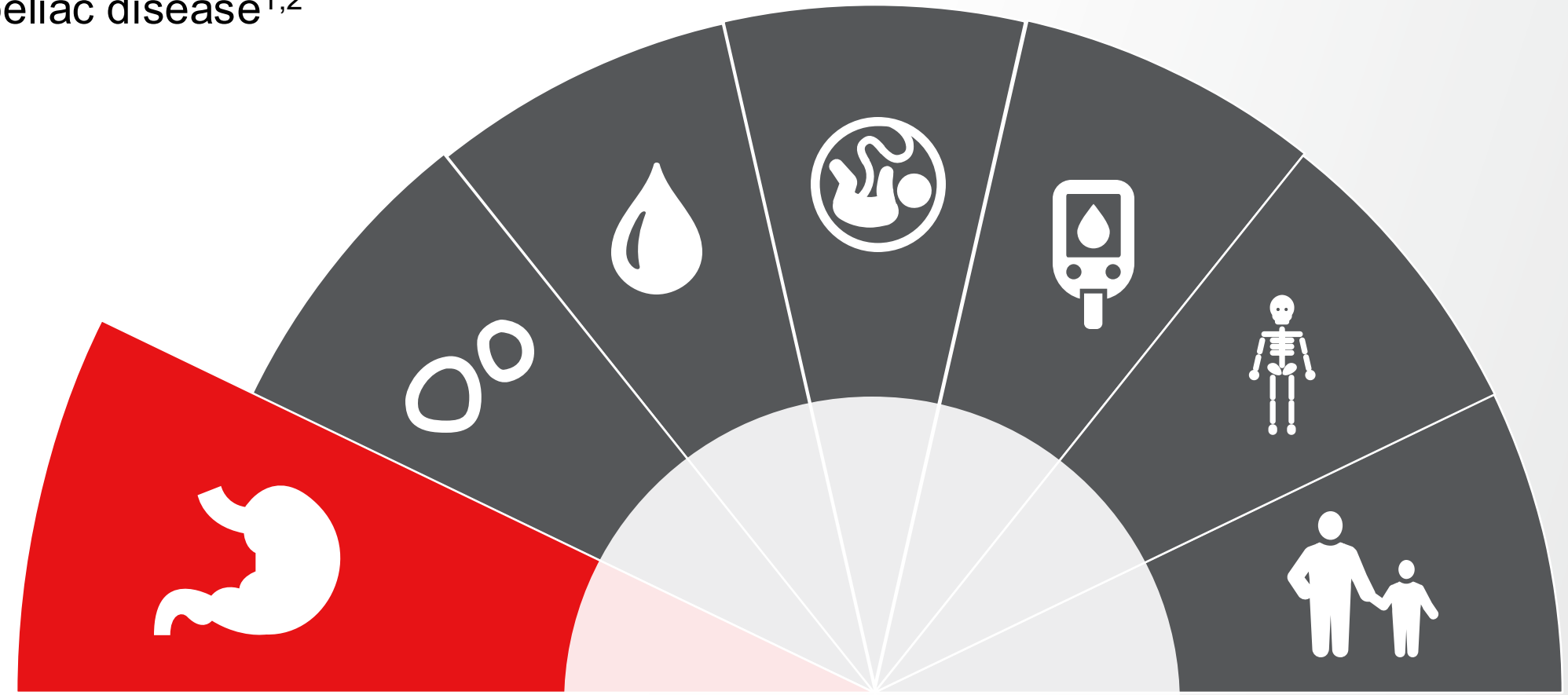
75% of coeliac disease cases in the EU population remain undiagnosed

Several guidelines recommend coeliac disease screening in high risk groups[1]

Who should be tested: identifying high risk groups

Irritable bowel syndrome

Up to 6% of IBS patients have coeliac disease[1,2]



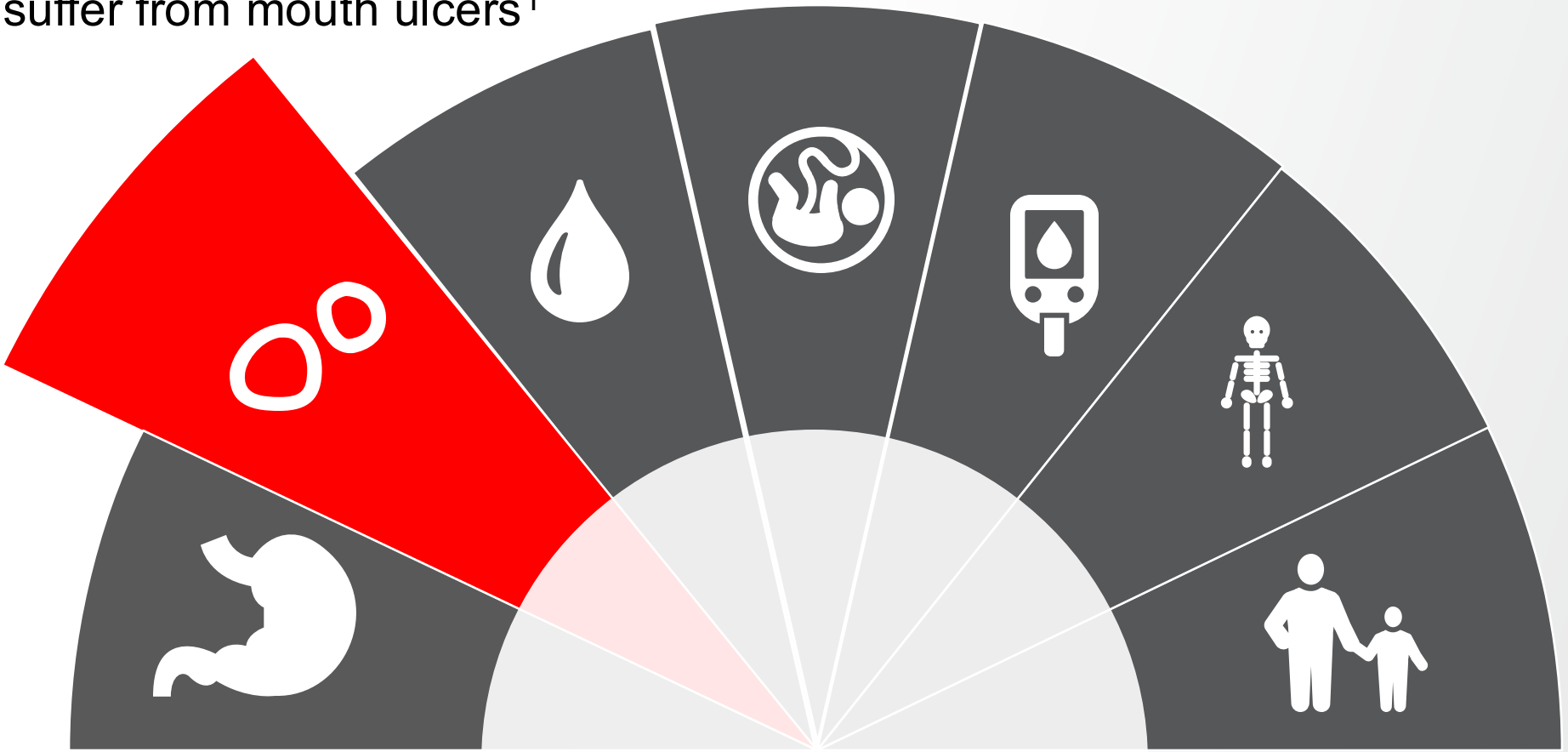
1.El-Salhy M, et al. The relation between celiac disease, nonceliac gluten sensitivity and irritable bowel syndrome. Nutr J. 2015;14:92.

2. Shiha MG, et al. Global prevalence of celiac disease in patients with Rome III and Rome IV irritable bowel syndrome: a systematic review and meta-analysis. *Am J Gastroenterol.* 2025;120(12):2776–2787.

Who should be tested: identifying high risk groups

Stomatitis

Up to 34.6% of patients with coeliac disease suffer from mouth ulcers[1]

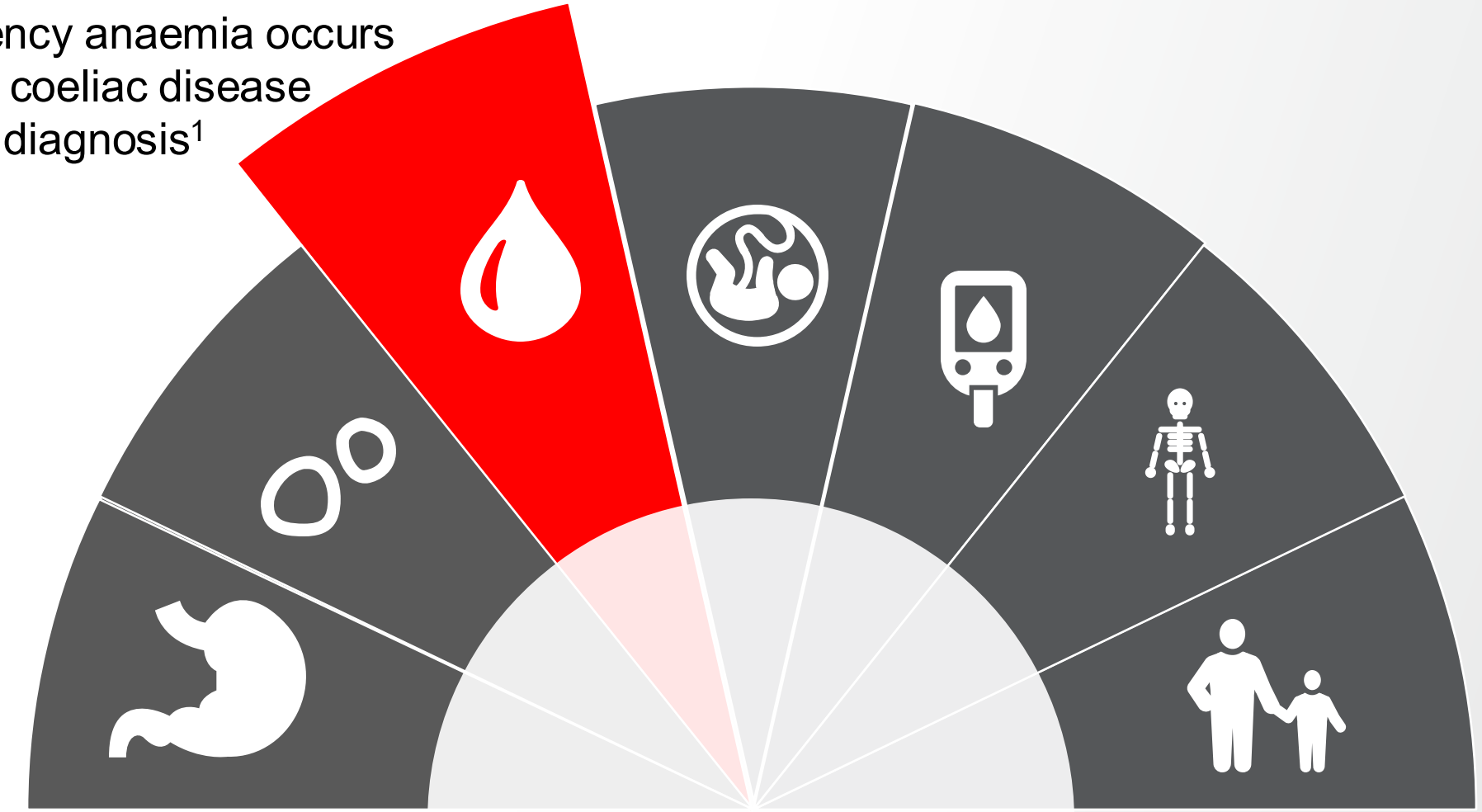


1. Lucchese A, et al. Beyond the gut: A systematic review of oral manifestations in celiac disease. *J Clin Med*. 2023;12:3874.

Who should be tested: identifying high risk groups

Unexplained anaemia

Iron deficiency anaemia occurs in >50% of coeliac disease patients at diagnosis[1]

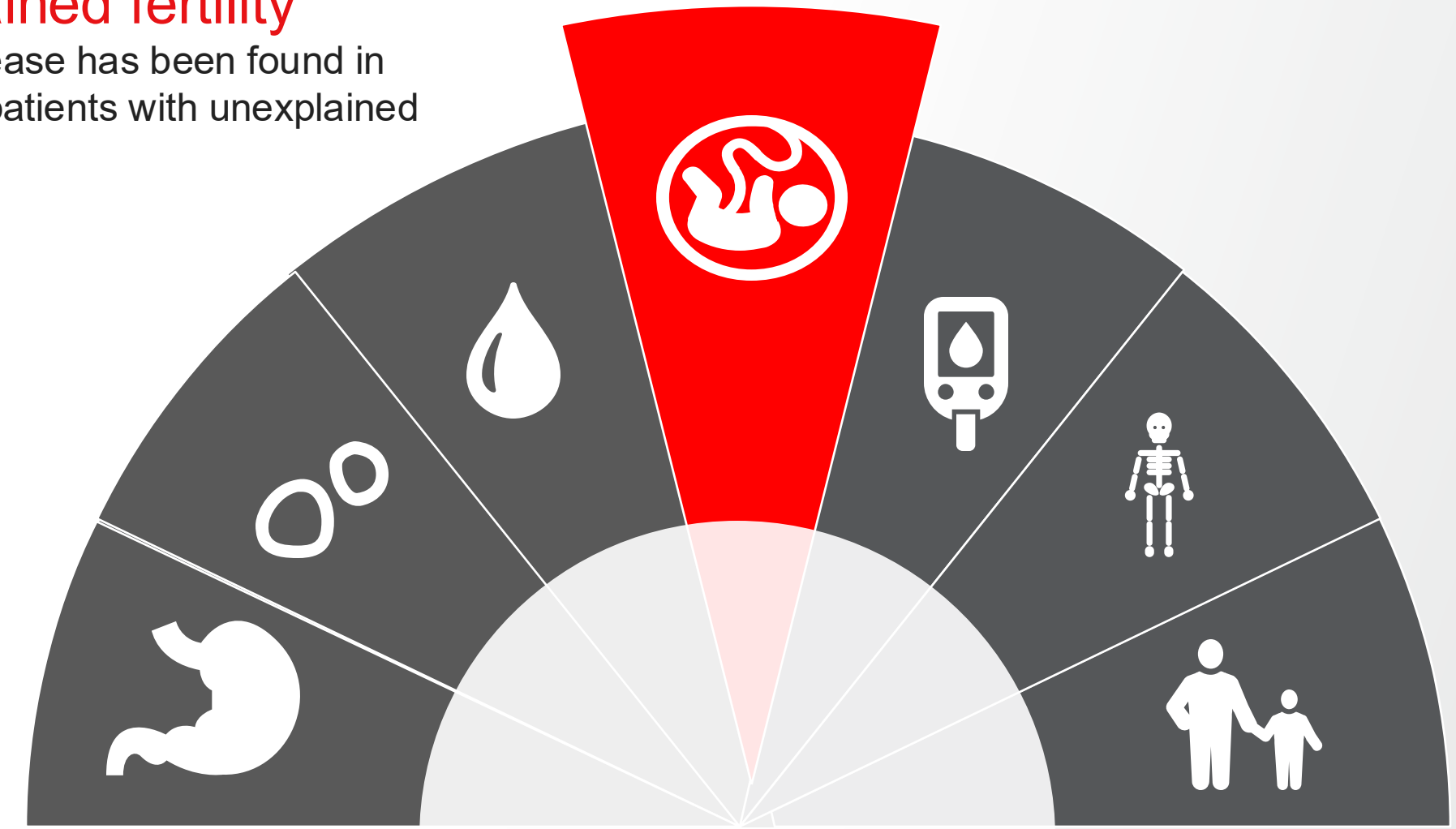


1. Seidita A, et al. Anemia in celiac disease: Prevalence, associated clinical and laboratory features, and persistence after gluten-free diet. *J Pers Med*. 2022;12:1582.

Who should be tested: identifying high risk groups

Unexplained fertility

Coeliac disease has been found in **4 to 8%** of patients with unexplained infertility[1]

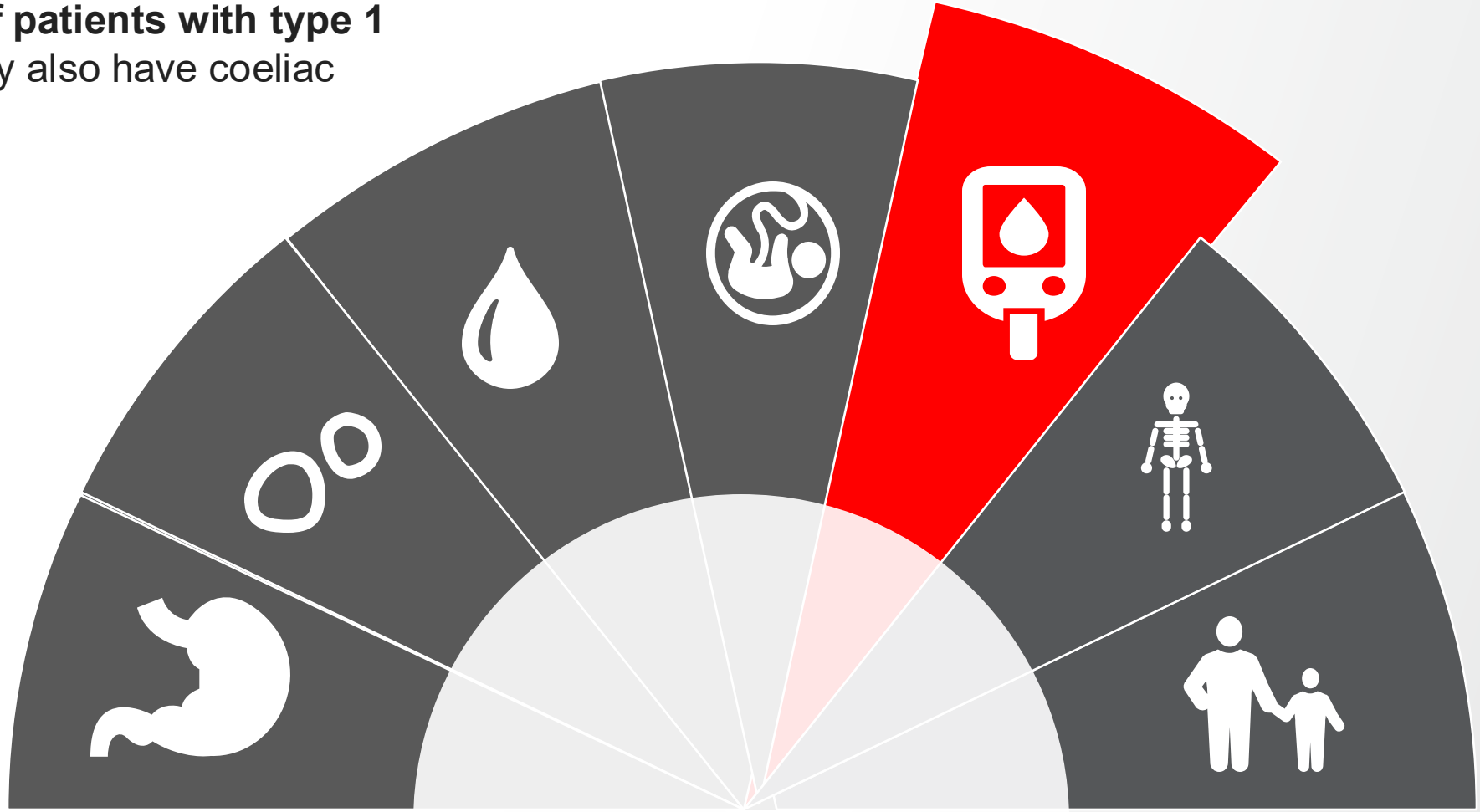


1. Di Simone N, et al. Celiac disease and reproductive failures: An update on pathogenic mechanisms. Am J Reprod Immunol. 2021;85:e13334.

Who should be tested: identifying high risk groups

Type 1 diabetes mellitus

Up to **16% of patients with type 1 diabetes** may also have coeliac disease[1,2]

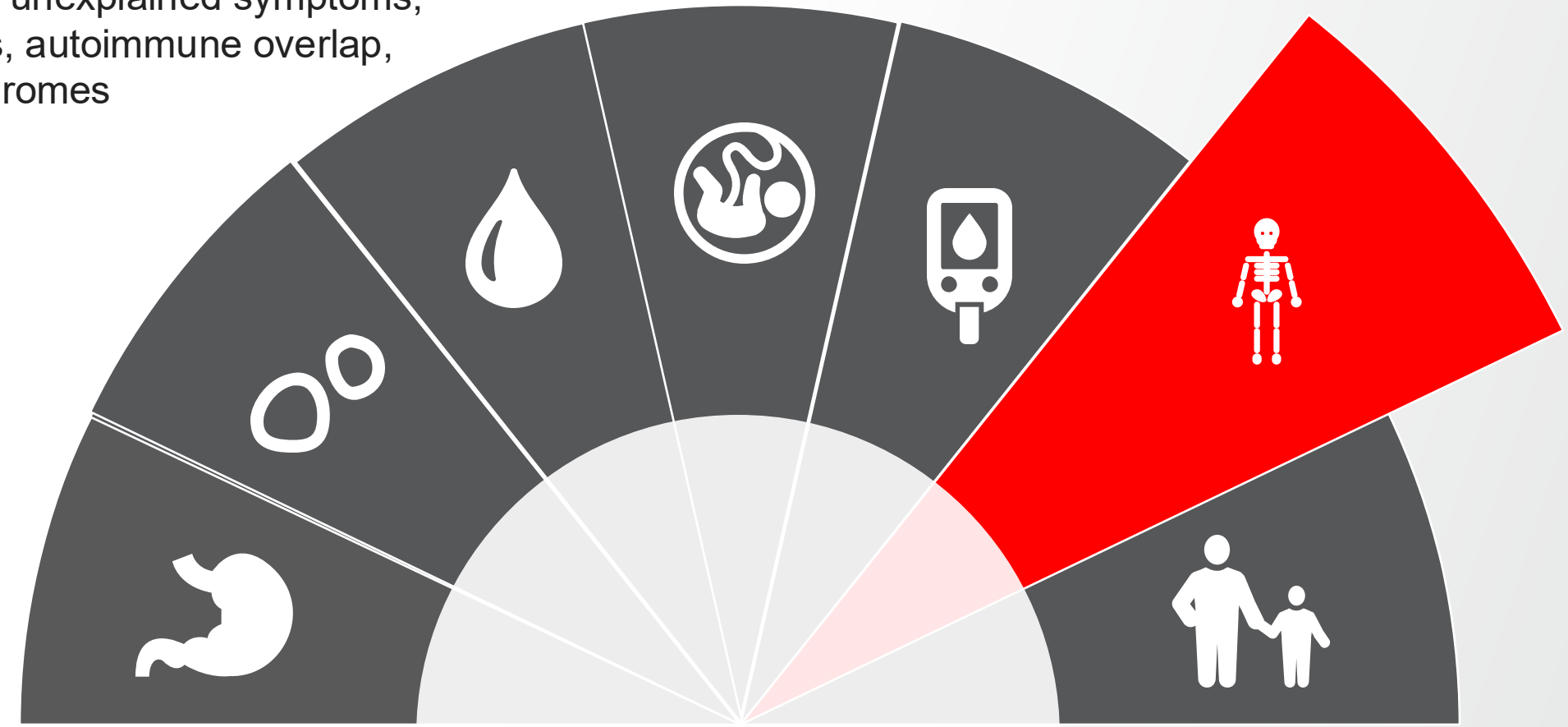


1. Arora S, et al. Unveiling the link: A comprehensive narrative review of the relationship between type 1 diabetes mellitus and celiac disease. *Cureus*. 2023;15(10):e47726.
2. Elfström P, Sundström J, Ludvigsson JF. Systematic review with meta-analysis: Associations between coeliac disease and type 1 diabetes. Aliment Pharmacol Ther. 2014;40:1123–1132.

Who should be tested: identifying high risk groups[1,2]

Other risk groups

Guidelines recommend testing in primary care for patients with unexplained symptoms, osteoporosis, autoimmune overlap, genetic syndromes

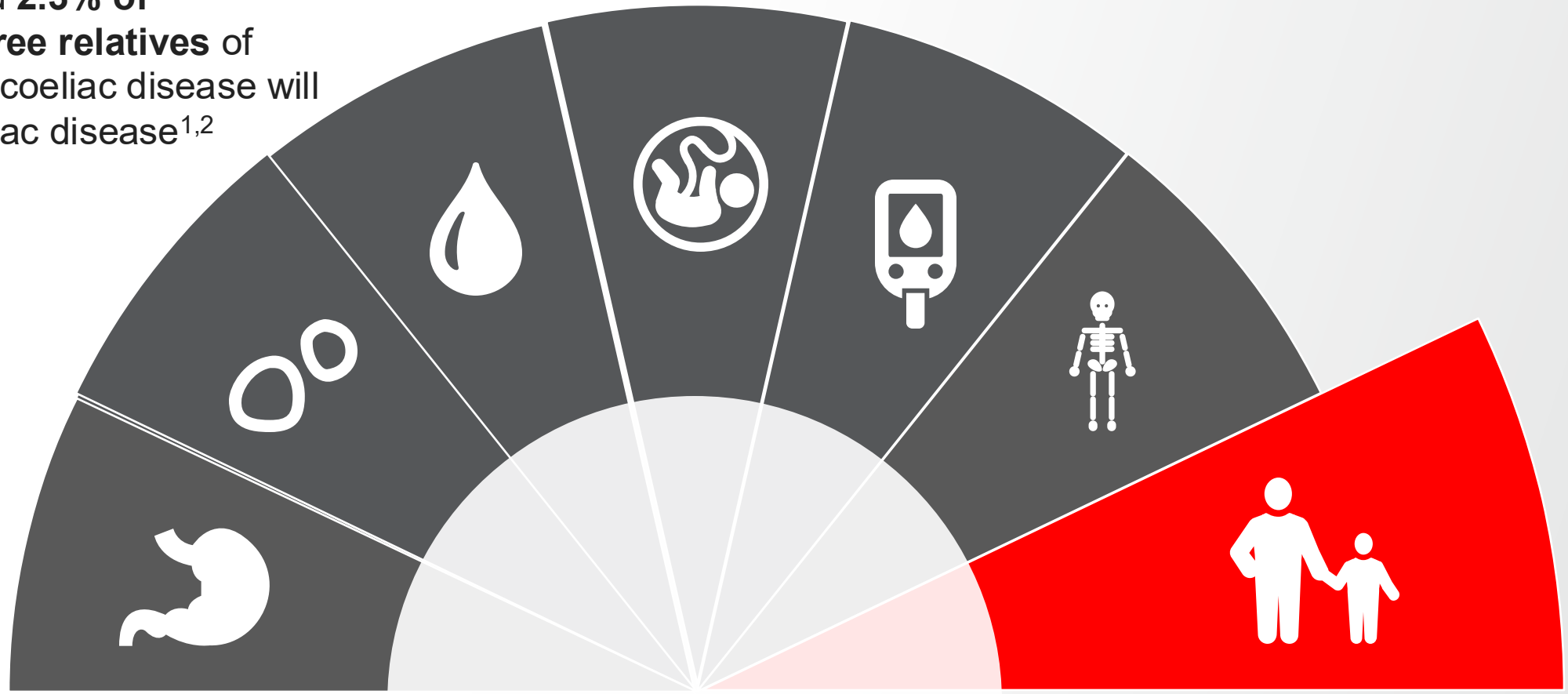


1. National Institute for Health and Care Excellence (NICE). Coeliac disease: recognition, assessment and management (NG20*)*. London: NICE; 2015.
2. Husby S, et al. European Society for Pediatric Gastroenterology, Hepatology, and Nutrition guidelines for diagnosing coeliac disease 2020. J Pediatr Gastroenterol Nutr. 2020;70(1):141–156.

Who should be tested: identifying high risk groups[1,2]

First degree relatives

Up to 11% of first-degree relatives and **2.3% of second-degree relatives** of patients with coeliac disease will develop coeliac disease[1,2]



1. Singh P, et al. Risk of celiac disease in the first- and second-degree relatives of patients with celiac disease: A systematic review and meta-analysis. *Am J Gastroenterol*. 2015;110:1539–1548.
2. Karimzadhagh S, et al. Global prevalence and clinical manifestations of celiac disease among first-degree relatives: A systematic review and meta-analysis. Am J Gastroenterol. 2024.



Guidelines for the diagnosis and management of coeliac disease

Primary care approach when suspecting coeliac disease[1,2]

Step 1: Accurate test request

1. Patient must be following a gluten containing diet
2. Start to test for tTG IgA plus total IgA

Step 2: Test interpretation

1. tTG IgA negative + normal IgA :
   → Coeliac disease unlikely

2. tTG IgA negative + low IgA :
   → Possible IgA deficiency
   IgG-based tests should be performed

3. tTG IgA positive
   → Coeliac disease likely
   → Refer to gastroenterologist
   (biopsy or no-biopsy pathway per guidelines)

Step 3: Lead within scope

- Primary care: identify and refer correctly
- Do not start gluten-free diet before diagnosis
- Consider testing in high-risk groups

tTG IgA testing is available as the EliA™ Celikey IgA assay on the Phadia™ instruments.

1. Husby S, et al. European Society Paediatric Gastroenterology, Hepatology and Nutrition guidelines for diagnosing coeliac disease. J Pediatr Gastroenterol Nutr. 2020;70(1):141-156.
2. Al-Toma A, et al. European Society for the Study of Coeliac Disease 2025 Updated guidelines on the diagnosis and management of coeliac disease in adults. United European Gastroenterol J. 2025;00:1–32.

Coeliac disease: confirmation biopsy approach

Antibodies against tissue transglutaminase (EliA Celikey™ IgA test) + total IgA

EliA Celikey™ IgA test is negative **and** total IgA normal

EliA Celikey™ IgA test is negative **and** total IgA low

Consider possible IgA deficiency

EliA Celikey™ IgG test or EliA™ Gliadin^{DP} IgG test

EliA Celikey™ IgG test **and** EliA Gliadin^{DP}™ IgG test are negative

EliA Celikey™ IgG test **or** EliA Gliadin^{DP}™ test is IgG positive

EliA Celikey™ IgA test is positive

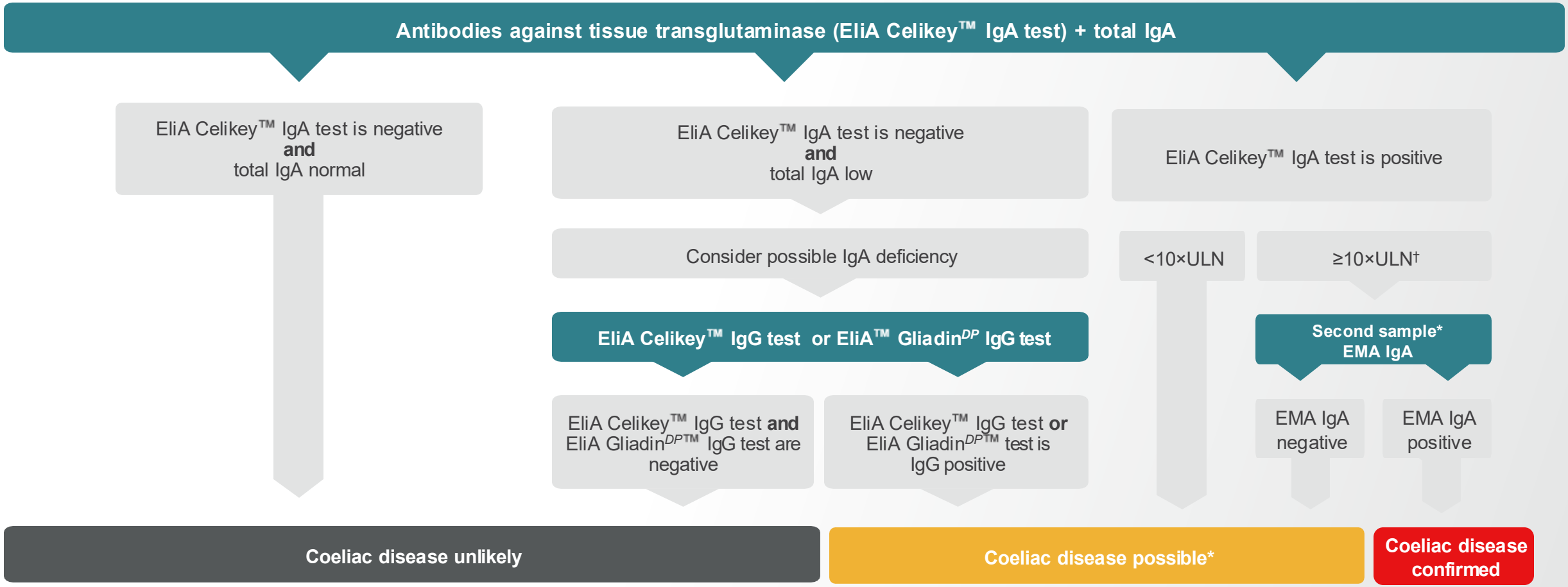
Coeliac disease unlikely

Coeliac disease likely*

* Investigation and confirmation by a specialist: ≥ 4 biopsies from distal duodenum and ≥ 1 biopsy from bulb

1. Husby S, et al. European Society Paediatric Gastroenterology, Hepatology and Nutrition guidelines for diagnosing coeliac disease. J Pediatr Gastroenterol Nutr. 2020;70(1):141-156.
2. Al-Toma A, et al. European Society for the Study of Coeliac Disease 2025 Updated guidelines on the diagnosis and management of coeliac disease in adults. United European Gastroenterol J. 2025;00:1–32.
3. Rubio-Tapia A, et al. American College of Gastroenterology guidelines update: diagnosis and management of celiac disease. Am J Gastroenterol. 2023;118(1):59-76.

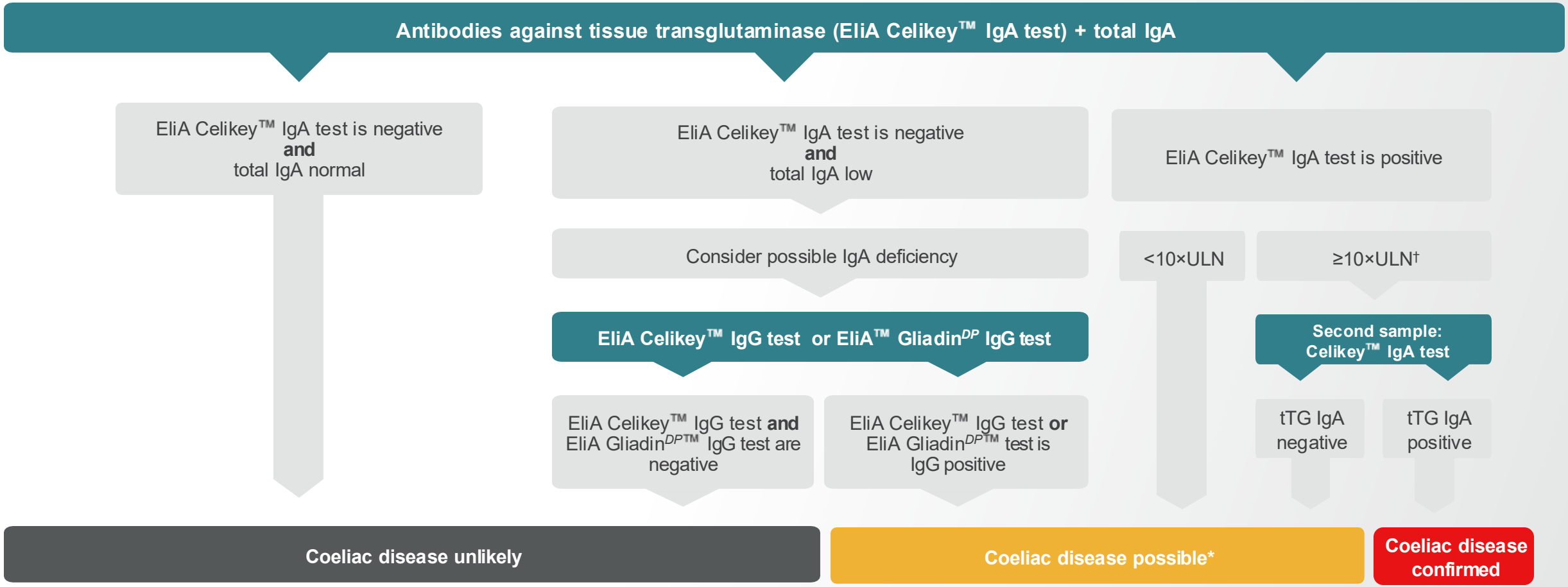
Coeliac disease: non-biopsy approach



* Investigation and confirmation by a specialist: ≥ 4 biopsies from distal duodenum and ≥ 1 biopsy from bulb

1. Husby S, et al. European Society Paediatric Gastroenterology, Hepatology and Nutrition guidelines for diagnosing coeliac disease. J Pediatr Gastroenterol Nutr. 2020;70(1):141-156.
2. Penny HA, et al. Accuracy of a no-biopsy approach for the diagnosis of coeliac disease across different adult cohorts. Gut. 2021;70(5):876-883.

Coeliac disease: non-biopsy approach by European Society for the Study of Coeliac Disease (ESsCD)

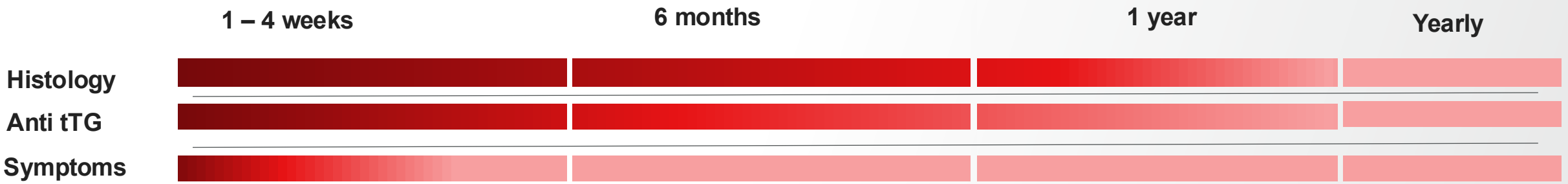


* Investigation and confirmation by a specialist: ≥ 4 biopsies from distal duodenum and ≥ 1 biopsy from bulb

1. Al-Toma A, et al. European Society for the Study of Coeliac Disease 2025 Updated guidelines on the diagnosis and management of coeliac disease in adults. United European Gastroenterol J. 2025;00:1–32..

Monitoring coeliac disease in adults[1]

Expected normalization timeline



No improvement | **Improvement**

Dietician referral; start GFD	Dietary adherence		Dietician if needed
Baseline laboratory testing	Repeat tTG IgA testing		tTG IgA testing if needed
Baseline clinical assessment	Symptom check	DEXA scan	Consider follow-up duodenal biopsy

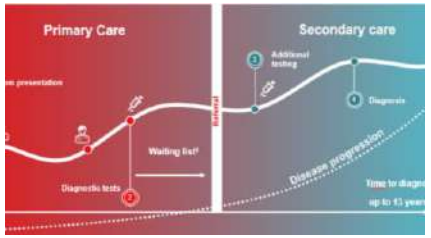
Ref 1. Elli L, et al. Guidelines for best practices in monitoring established coeliac disease in adult patients. Nat Rev Gastroenterol Hepatol. 2024;21(3):198-215



Summary

Your key take-aways

Consider coeliac disease early in the diagnostic process



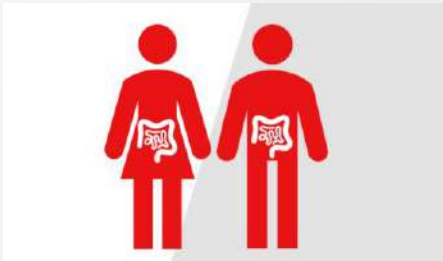
- Coeliac disease affects 1% of the general population
- Up to 12% of general practitioner's visits involve gastrointestinal symptoms
- Symptoms are often non-classical or extra-intestinal
- Diagnostic delay is common

Early diagnosis improves outcome



- Prevents long-term complications
- Reduces repeated consultations
- Decreases secondary care burden
- ESPGHAN and ESsCD provide clear testing pathways

Practical steps in primary care



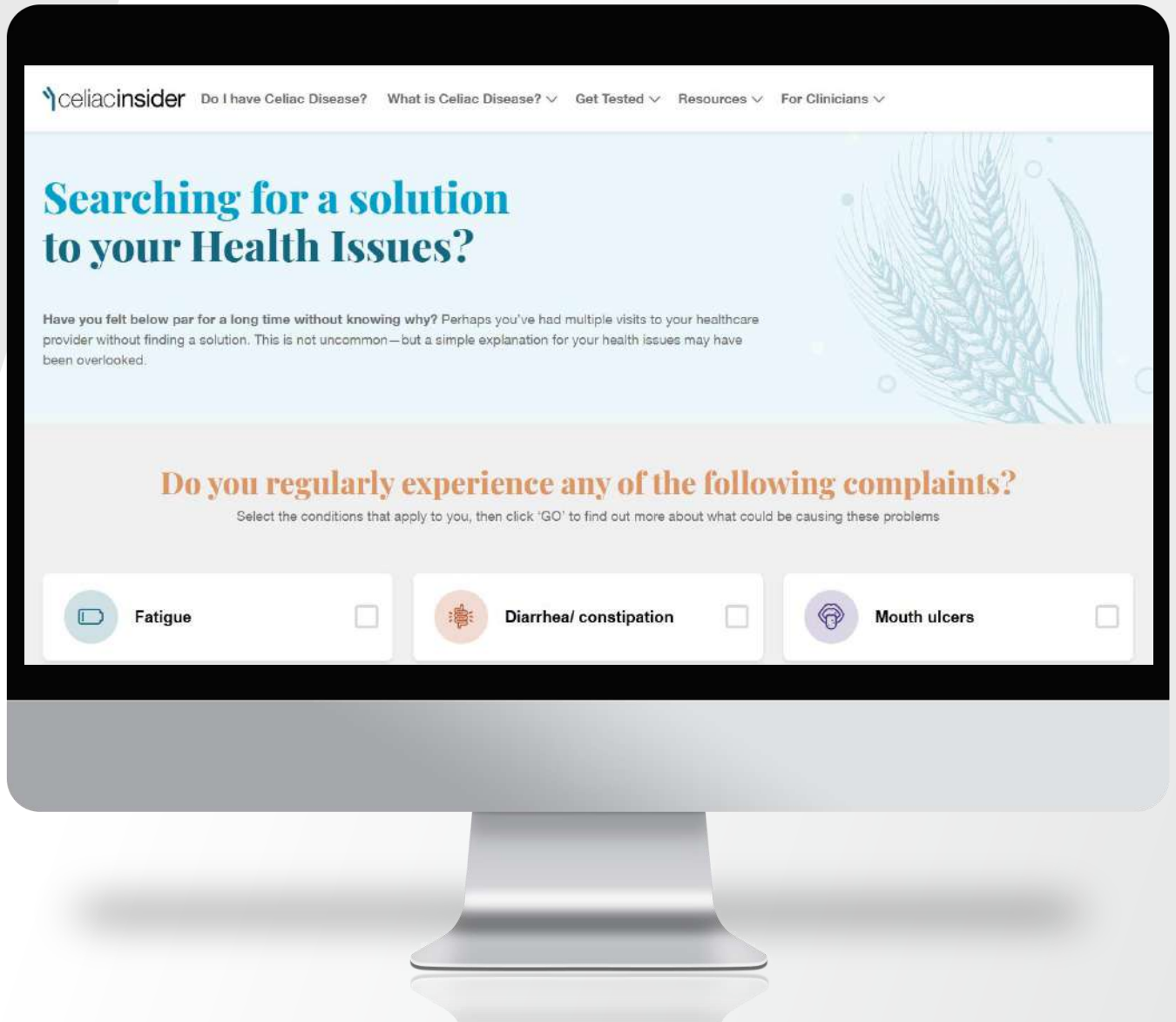
- Test while the patient is on a gluten containing diet
- Order: total IgA plus tTG IgA (first line)
- Refer positive cases to gastroenterology
- Provide patients with reliable education: Coeliac Insider

Think of coeliac disease — especially in persistent, unexplained or atypical presentations

Want to learn more about coeliac disease?

Coeliac Insider is an educational resource for patients and healthcare professionals.

Scan for
coeliacinsider.com





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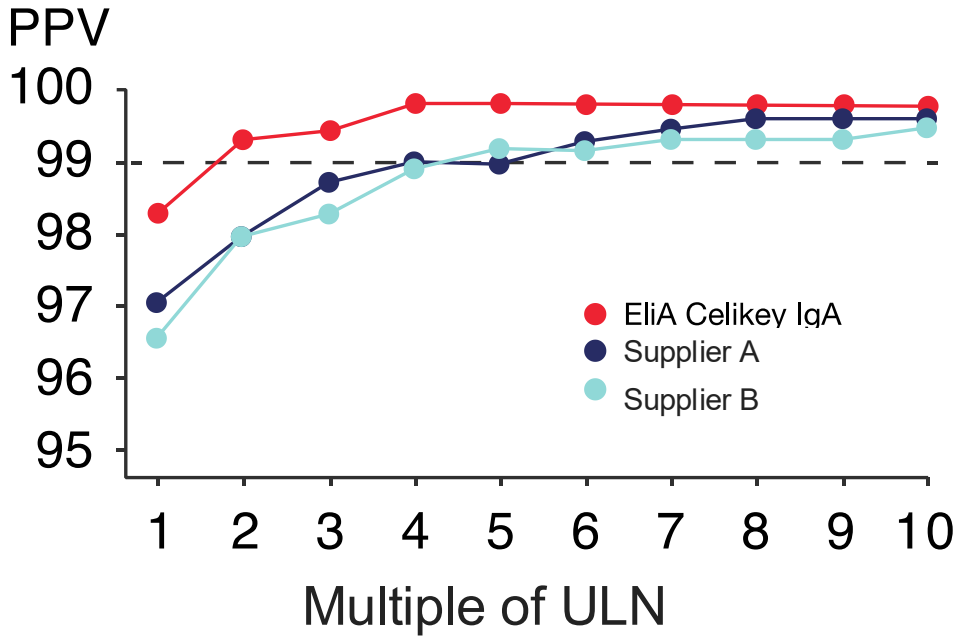
Additonal slides – for explanation of laboratory tests



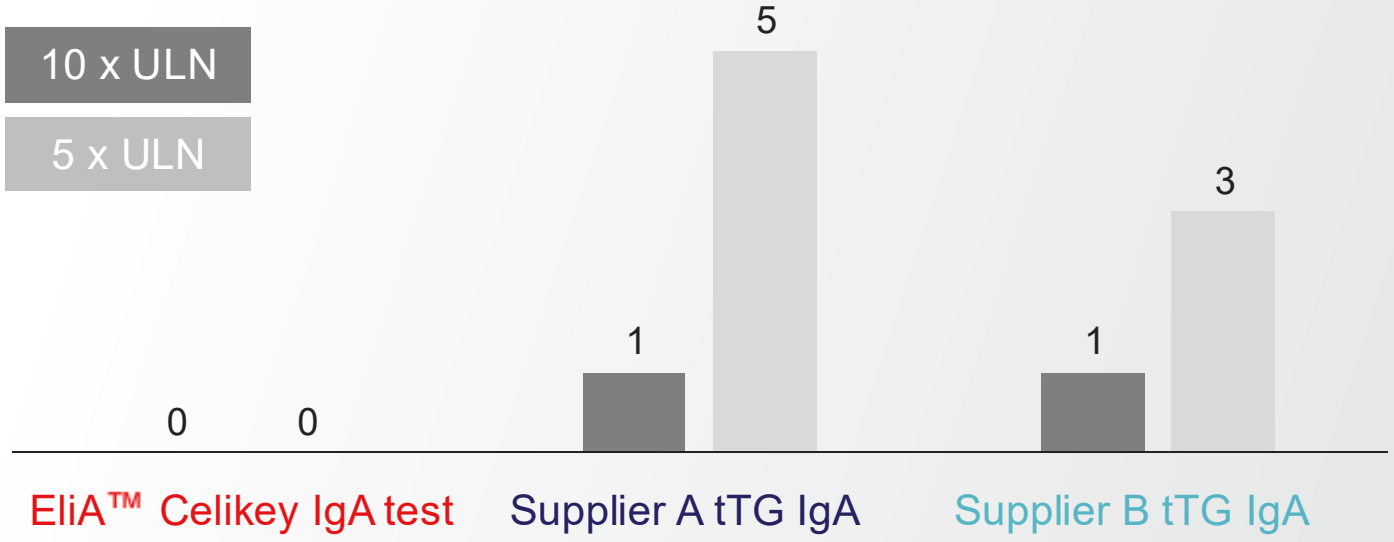
Understanding test performance

The importance of diagnostic accuracy - ProCEDe study[1]

Positive predictive value (PPV)



Number of false positive results at 5x and 10x ULN*



In published studies, EliA Celikey IgA test achieved a 99% PPV already at 2x ULN

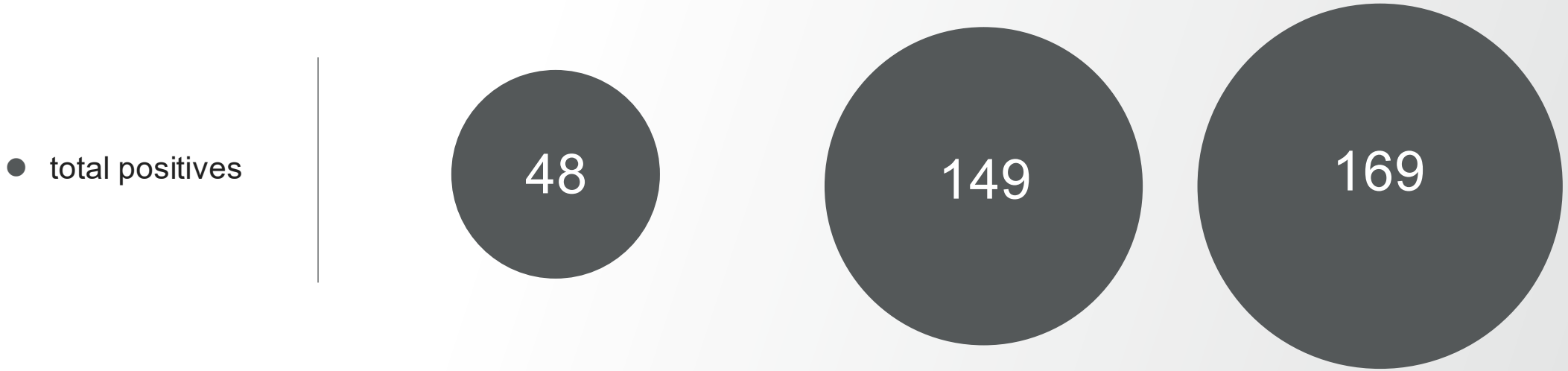
1. Werkstetter KJ, et al. Accuracy in Diagnosis of coeliac Disease Without Biopsies in Clinical Practice. Gastroenterology. 2017;153(4):924-35.

* ULN = upper limit of normal

The importance of diagnostic accuracy[1]

Leveraging real-world data for extrapolation: 5,000 children‘s samples, prevalence of 1%, 50 coeliac disease patients

	EliA™ Celikey IgA	Test 2	Test 3
Sensitivity	96.6%	100%	100%
Specificity	100%	98%	97.6%
Positive results	48	149	169
True positives			
False positives			

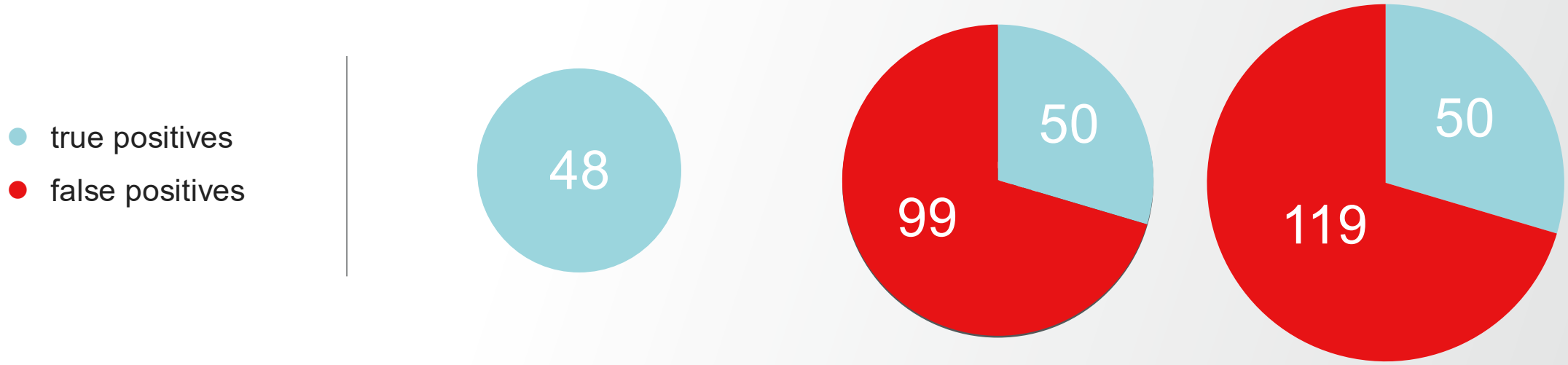


1. Casteleijn MG, et al. Diagnostic accuracy of various serological tests for celiac disease. Clin Chem Lab Med. 2022;60(12):2001-2009

The importance of diagnostic accuracy[1]

Leveraging real-world data for extrapolation: 5,000 children‘s samples, prevalence of 1%, 50 coeliac disease patients

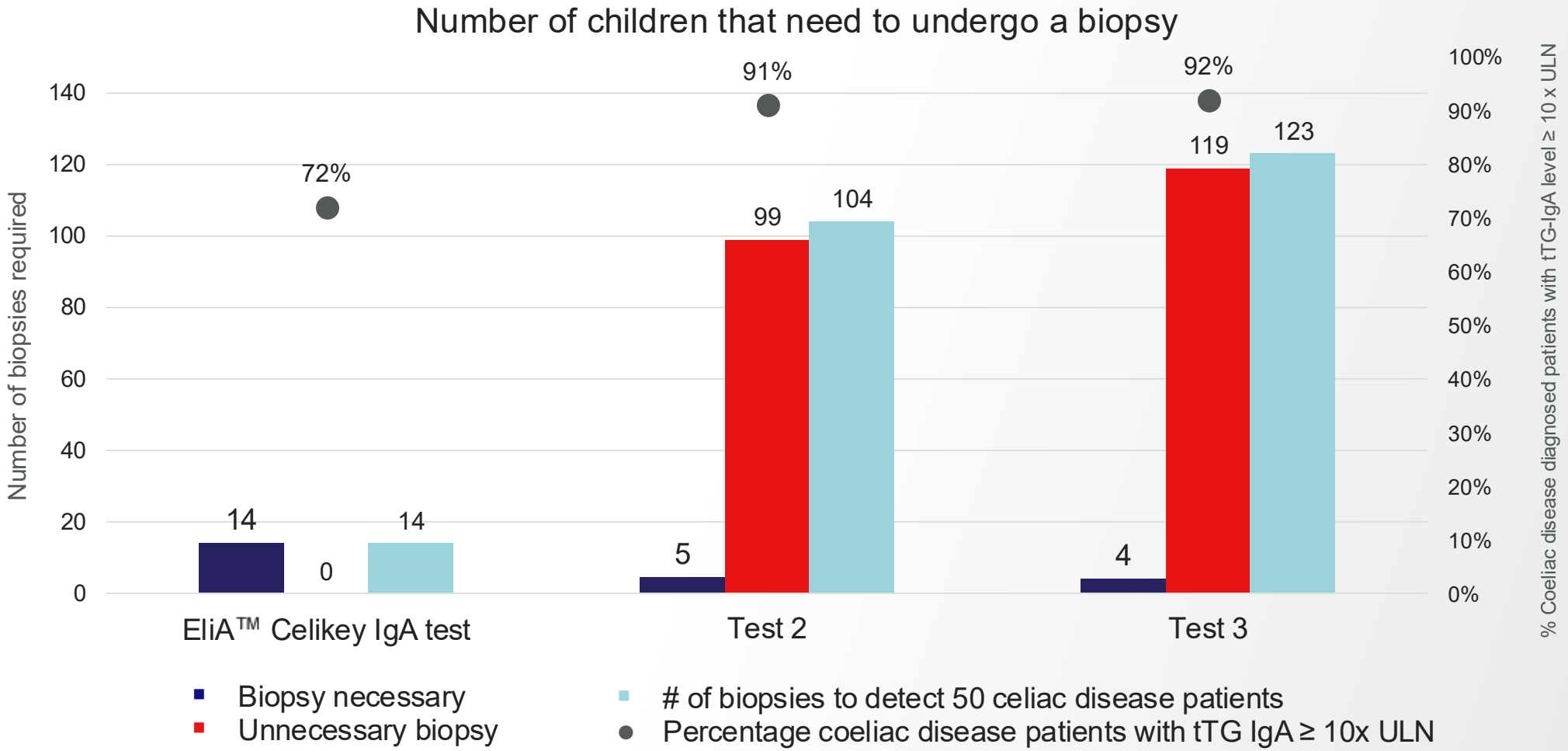
	EliA™ Celikey IgA	Test 2	Test 3
Sensitivity	96.6%	100%	100%
Specificity	100%	98%	97.6%
Positive results	48	149	169
True positives	48	50	50
False positives	0	99	119



1. Casteleijn MG, et al. Diagnostic accuracy of various serological tests for celiac disease. Clin Chem Lab Med. 2022;60(12):2001-2009

Accurate testing results in fewer biopsies[1]

Leveraging real-world data for extrapolation: 5,000 children's samples, prevalence of 1%, 50 coeliac disease patients



1. Casteleijn MG, et al. Diagnostic accuracy of various serological tests for celiac disease. Clin Chem Lab Med. 2022;60(12):2001-2009