Turner syndrome

Bibliographic reference	**Bonamico et al. (2002)**
Study type	Cross-sectional survey
Study quality	The Joanna Briggs Institute Prevalence Critical Appraisal Tool (http://ijhpm.com/article_2870_607.html) 1. Was the sample representative of the target population? YES 2. Were study participants recruited in an appropriate way? NO – Unclear is consecutive sample recruited 3. Was the sample size adequate? YES 4. Were the study subjects and the setting described in detail? YES 5. Was the data analysis conducted with sufficient coverage of the identified sample? YES 6. Were objective, standard criteria used for the measurement of the condition? YES 7. Was the condition measured reliably? YES 8. Was there appropriate statistical analysis? YES 9. Are all important confounding factors/subgroups/differences identified and accounted for? YES 10. Were subpopulations identified using objective criteria? NA Overall risk of bias = MODERATE
Country	Italy
Number of patients	N=389
Study population	Inclusion: patients with Turner syndrome enrolled by the Italian Society of Pediatric Gastroenterology and Hepatology and the TS Italian Study Group from various centres of the northern, central, southern, and insular regions, making the sample fairly representative of the whole population, age range 7 to 38yrs
Details of coeliac testing	IgA AGA and/or EMA and biopsies
Results	N=25 (6.4%) diagnosed with coeliac disease N=10 (40%) classic form of coeliac disease, N=8 (32%) atypical, N=7 (28%) silent
Source of funding	Consiglio Nazionale delle Ricerche Grant
Conflicts of interest	
Comments	

Definitions of abbreviations are given at the end of this document.

Bibliographic reference	**Dias et al. (2010)**
Study type	Cross-sectional survey
Study quality	The Joanna Briggs Institute Prevalence Critical Appraisal Tool (http://ijhpm.com/article_2870_607.html) 1. Was the sample representative of the target population? YES 2. Were study participants recruited in an appropriate way? NO – Unclear is consecutive sample recruited 3. Was the sample size adequate? YES 4. Were the study subjects and the setting described in detail? YES 5. Was the data analysis conducted with sufficient coverage of the identified sample? YES 6. Were objective, standard criteria used for the measurement of the condition? YES 7. Was the condition measured reliably? YES 8. Was there appropriate statistical analysis? YES 9. Are all important confounding factors/subgroups/differences identified and accounted for? YES 10. Were subpopulations identified using objective criteria? NA Overall risk of bias = MODERATE
Country	Brazil
Number of patients	N=56 women with Turner Syndrome
Study population	Inclusion: women with Turner syndrome confirmed on cytogenetic testing who were followed at a Clinical Genetic Unit a t a hospital and who were on a gluten-containing diet and without a prior diagnosis of CD Mean age at diagnosis: 5.5 ± 4.4 years Mean age at CD screening: 17.0 ± 9.3 years (from 10 month to 52 years)
Control	none
Length of follow-up	n/a
Details of coeliac testing	IgA levels (<5 mg/dL were considered abnormal) IgA EMA (distal portion of monkey oesophagus used as antigenic substrate, Inova Diagnostics, and fluorescein-labelled goat antibody as second substrate); confirmation with ELISA, Inova Diagnostics Biopsy if positive serology (characterised with Marsh criteria)
Results	3.6% (2/56) had biopsy-confirmed CD (both had positive IgA-EMA and IgA-tTG)
Source of funding	Celiac disease Investigation laboratory, Department of Paediatrics, University of Brasilia School of Mediciine, Brasilia

Conflicts of interest	Paper reports no conflicts of interest concerning this research
Comments	

Definitions of abbreviations are given at the end of this document.

Bibliographic reference	**Frost et al. (2009)**
Study type	Cross-sectional survey
Study quality	The Joanna Briggs Institute Prevalence Critical Appraisal Tool (http://ijhpm.com/article_2870_607.html) 1. Was the sample representative of the target population? YES 2. Were study participants recruited in an appropriate way? YES (consecutive sample recruited) 3. Was the sample size adequate? YES 4. Were the study subjects and the setting described in detail? YES 5. Was the data analysis conducted with sufficient coverage of the identified sample? YES 6. Were objective, standard criteria used for the measurement of the condition? YES 7. Was the condition measured reliably? YES 8. Was there appropriate statistical analysis? YES 9. Are all important confounding factors/subgroups/differences identified and accounted for? YES 10. Were subpopulations identified using objective criteria? NA Overall risk of bias = LOW
Country	UK
Number of patients	N=256 women with Turner Syndrome
Study population	Inclusion: consecutive women with karyotypically proven Turner syndrome attending an Adults Turner clinic as part of a health surveillance programme Median 29 years old (range 16 to 61)
Control	none
Length of follow-up	n/a
Details of coeliac testing	EMA using indirect immunofluorescence analysis with commercially available fixed sections of monkey oesophagus (Biodiagnostics Ltd, Worcestershire UK) as antigen substrate; diluted to 1:10) EMA IgA was detected with FITC-labelled sheep anti-human IgA conjugate (Dako, Ltd, Ely, UK) All positive patients were offered duodenal biopsy

	HLA typing was also offered to patients with positive EMA serology or previous diagnosis of CD
Results	5 were diagnosed prior to transition to adult care following clinical presentation Of the 251 without pre-diagnosis of CD, **3.2% (8/251) were positive for EMA** (none had symptoms suggestive of CD) All but one patient who denied biopsy were tested for histological signs of CD: **Partial or total villous atrophy was present in 2.8% (7/251)**
Source of funding	No specific grants for this research were received (however, the work was undertaken at UCLH/UCL who received a proportion of funding from the DH's NIHR Biomedical Research Centres funding scheme
Conflicts of interest	Not reported
Comments	

Definitions of abbreviations are given at the end of this document.

Bibliographic reference	**Mortensen et al. (2009)**
Study type	Cross-sectional survey
Study quality	The Joanna Briggs Institute Prevalence Critical Appraisal Tool (http://ijhpm.com/article_2870_607.html) 1. Was the sample representative of the target population? YES 2. Were study participants recruited in an appropriate way? NO – Unclear is consecutive sample recruited 3. Was the sample size adequate? YES 4. Were the study subjects and the setting described in detail? YES 5. Was the data analysis conducted with sufficient coverage of the identified sample? YES 6. Were objective, standard criteria used for the measurement of the condition? YES 7. Was the condition measured reliably? YES 8. Was there appropriate statistical analysis? YES 9. Are all important confounding factors/subgroups/differences identified and accounted for? YES 10. Were subpopulations identified using objective criteria? NA Overall risk of bias = MODERATE
Country	Denmark
Number of patients	N=107 patients with Turner syndrome
Study population	Inclusion: Danish Turner syndrome patients from the National Society of Turner Contact Groups in Denmark (through advertisement), and a number of hospital; all had undergone chromosome analysis

	Median age 36.7 years (range 6 -60)
Control	None
Length of follow-up	n/a
Details of coeliac testing	Total IgA was measured IgA AGA and IgA anti-tTG If IgA deficiency, AGA and anti-tTG IgG were determined
Results	**Anti-tTG, AGA or both were present: 18% (19/106)** **In 2 CD was known previously and 3 received a CD diagnosis (overall prevalence of diagnosed CD: 4.7% [5/106])** 97% (103/106) had normal IgA serum range The four youngest patients did not have autoantibodies Those with positive antibodies (for any autoimmunity) were significantly older than those without (38.0 ± 13.5 vs 29.4 ± 13.0 years, p = 0.001)
Source of funding	Dronning Louise Børnehospitals Forskningsfond
Conflicts of interest	Paper reports that the authors declared no conflicts of interest
Comments	Study considered prevalence of a number of autoimmunities in Turner syndrome, but only those related to coeliac disease are presented here

Definitions of abbreviations are given at the end of this document.