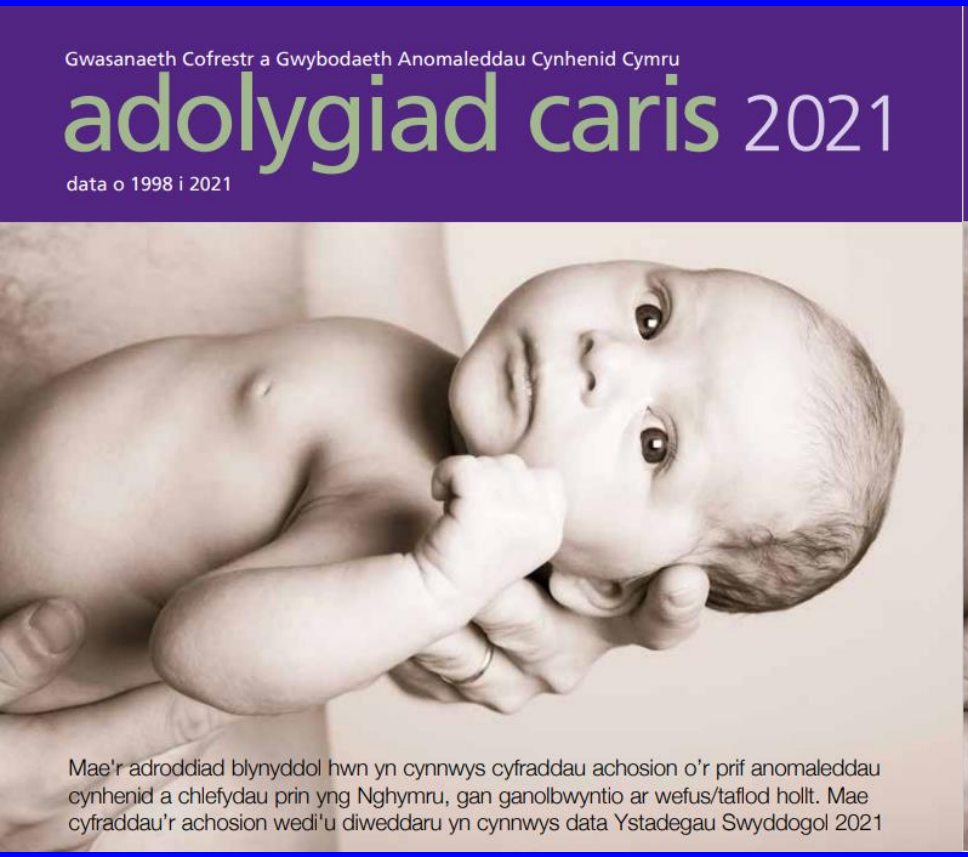
Diweddariad CARIS – Tachwedd 2022
CARIS Update – November 2022



Llion Davies

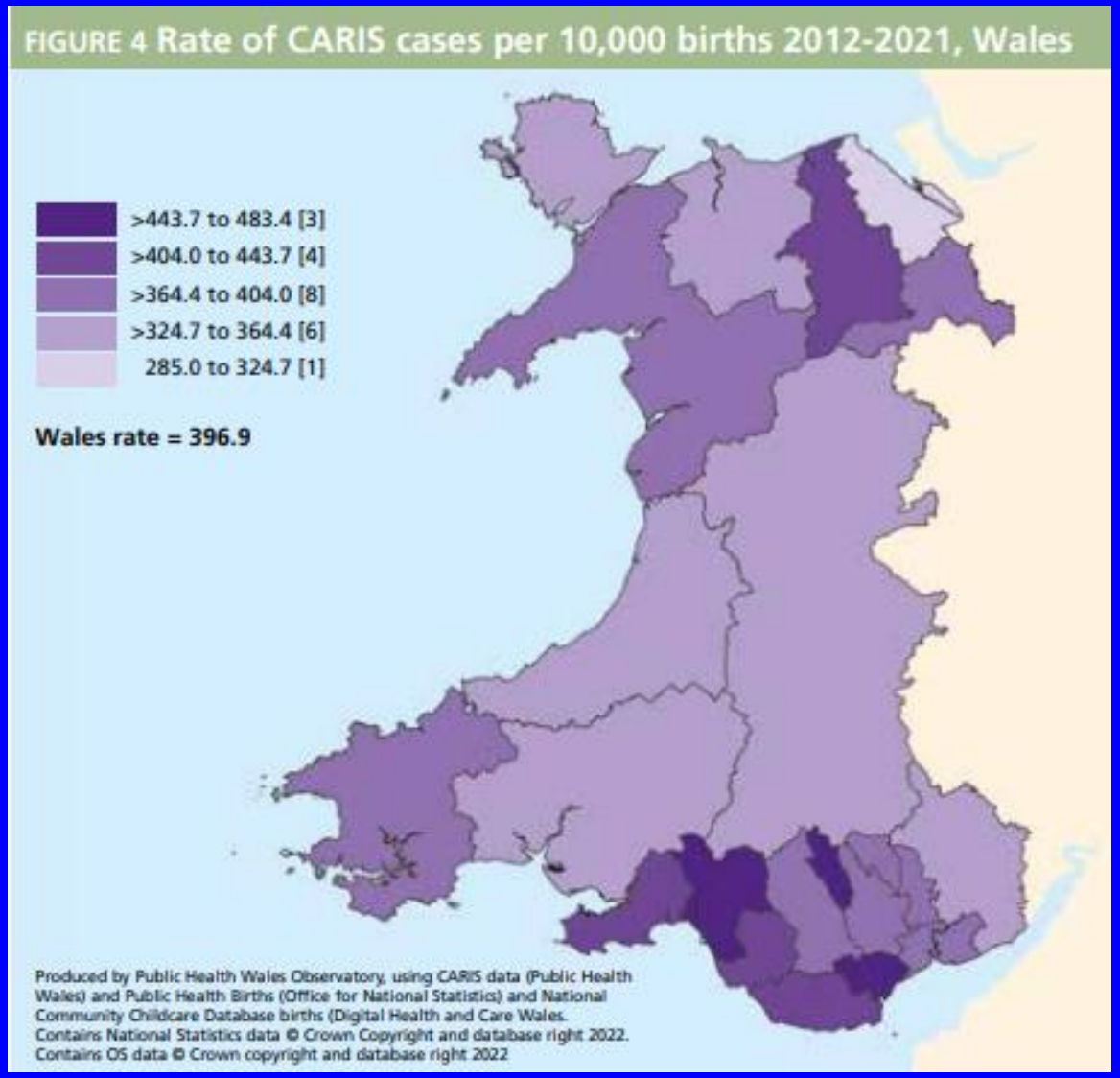
Updates

- Congenital Anomalies Headlines
- Cleft Lip and Palate
- Childhood Rare Diseases
- Adult Rare Diseases

Congenital Anomalies

- 38,528 cases since 1998
- 4.9% of all live and still births
- 85% liveborn
- 96.6% surviving to 1 year
- Single anomaly 57.7%
- Underlying chromosomal disorder 13.9%

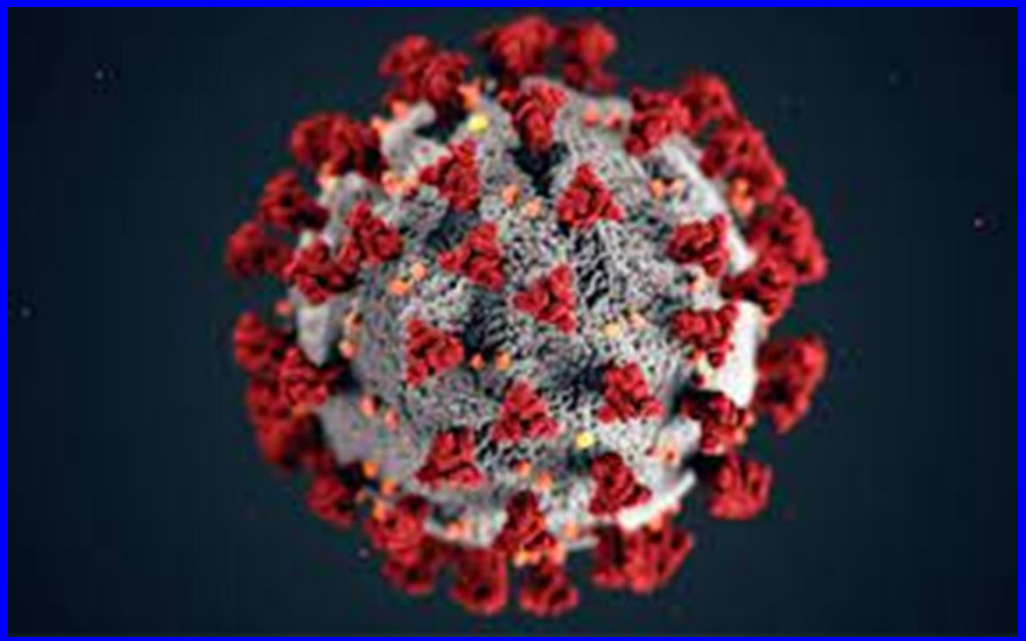
Geographical Reporting



- Similar to 2018
- Change to 10 year

COVID Impact

- Usual to have some lag
- Numbers predicted to raise
- COVID exacerbated for 2021
- Surgical data



Cleft Lip and Palate

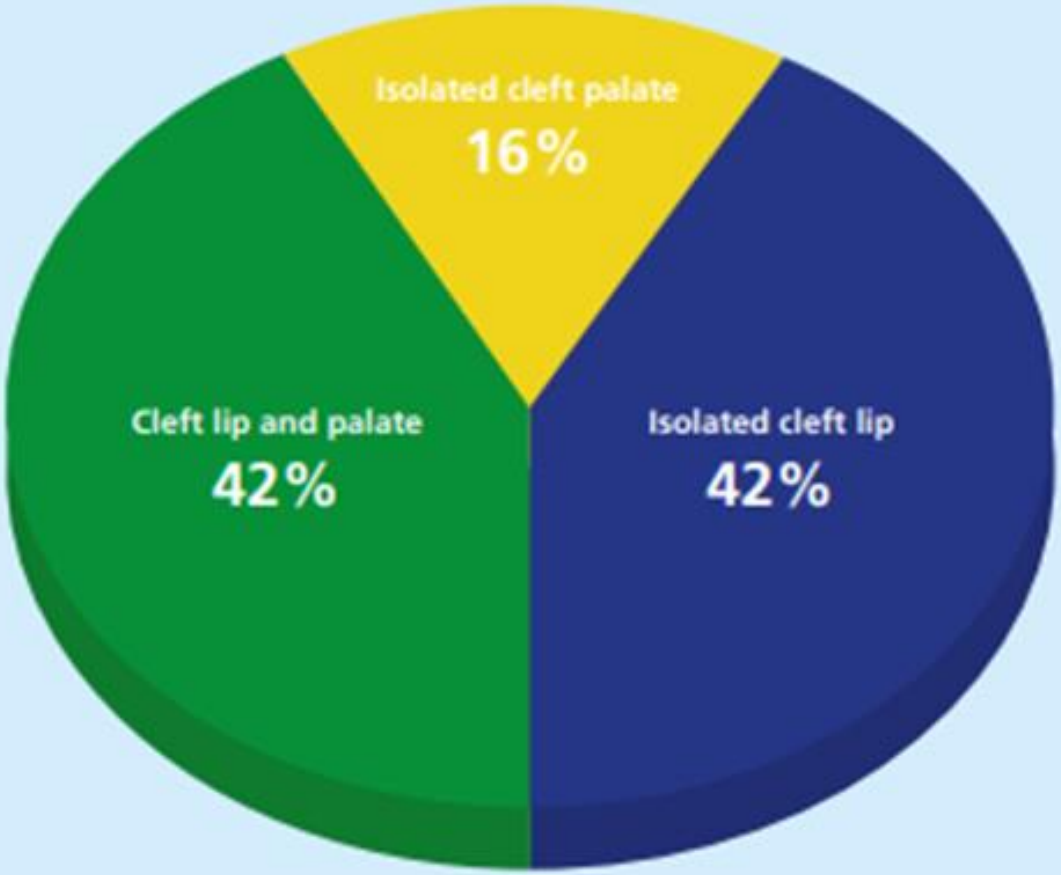
Cases with cleft lip/palate and/or Pierre Robin sequence, rate per 10,000 total births and percentage of cases live born, Wales, 1998-2000 to 2019-2021

Anomaly	Total cases	Average cases per year	Rate	% of cases liveborn	Trend (3 year rolling rate)
All cleft lip and palate	1,802	75	22.9	84.2%	
Isolated cleft palate	753	31	9.6	84.9%	
Isolated cleft lip	295	12	3.7	88.1%	
Cleft lip with or without cleft palate	897	37	11.4	81.0%	
Cleft lip and/or cleft palate	1,650	69	21.0	82.8%	
Pierre Robin sequence	152	6	1.9	100.0%	

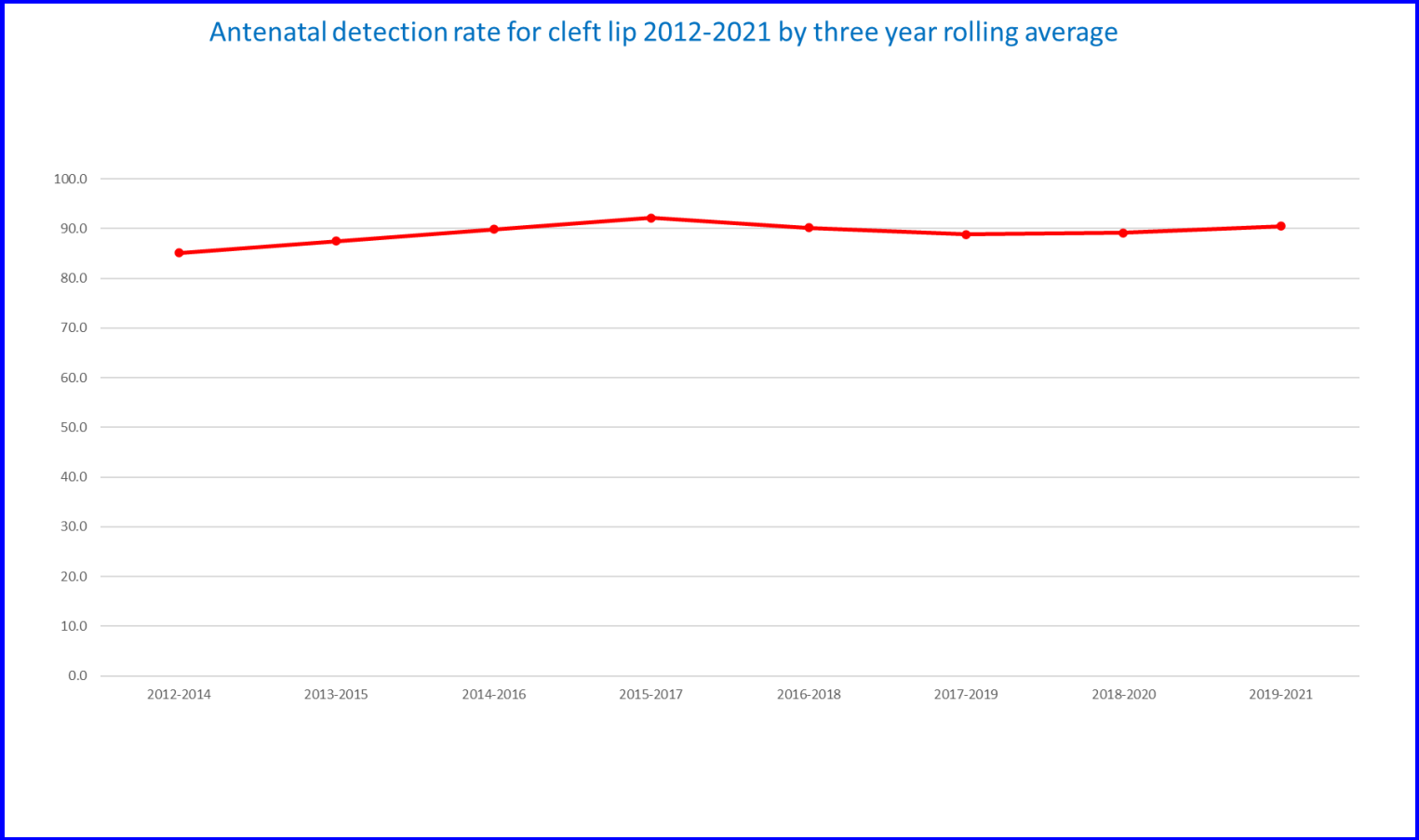
Produced by Public Health Wales Observatory, using CARIS, PHB (ONS) & NCCHD

Proportion Breakdown

FIGURE 2 Cleft lip/palate profile in Wales n=1802



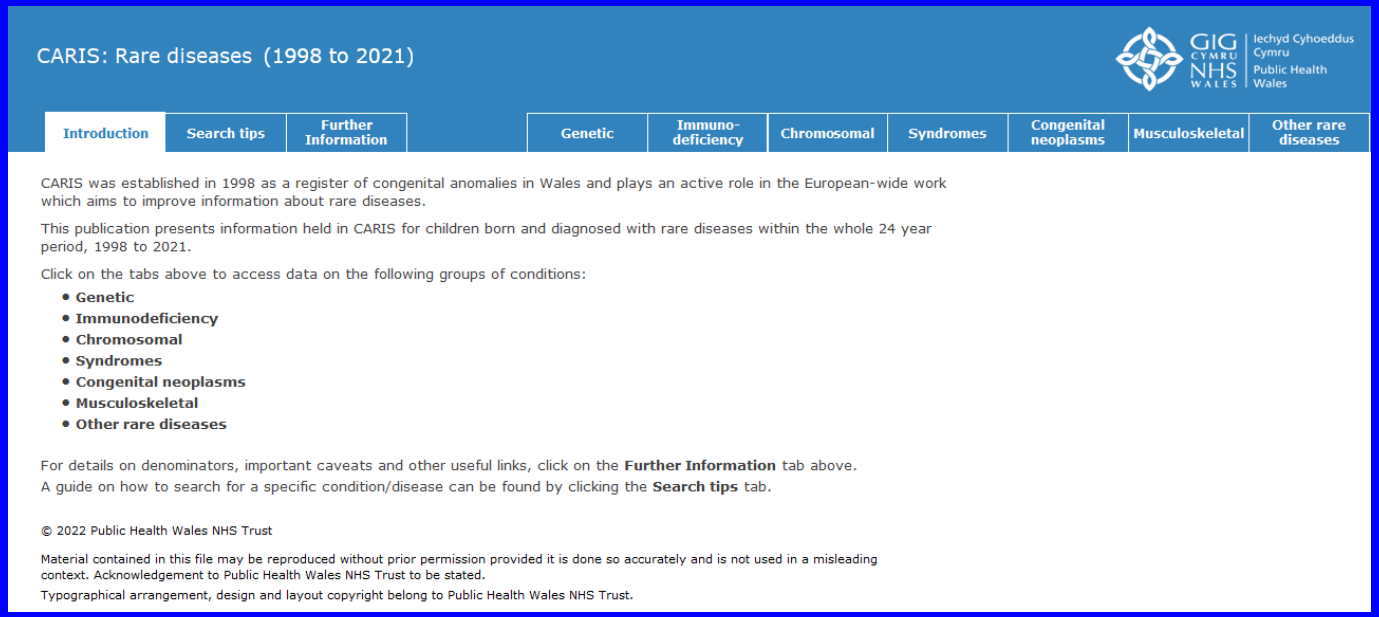
Isolated Cleft Lip



- **Isolated cleft palate:**
  - **?Geographical Variation**
  - **Deep dive to follow**

Childhood Rare Diseases

Official Stats



CARIS: Rare diseases (1998 to 2021)

GIG CYMRU NHS WALES | Iechyd Cyhoeddus Cymru Public Health Wales

Introduction | Search tips | Further Information | Genetic | Immuno-deficiency | Chromosomal | Syndromes | Congenital neoplasms | Musculoskeletal | Other rare diseases

CARIS was established in 1998 as a register of congenital anomalies in Wales and plays an active role in the European-wide work which aims to improve information about rare diseases.

This publication presents information held in CARIS for children born and diagnosed with rare diseases within the whole 24 year period, 1998 to 2021.

Click on the tabs above to access data on the following groups of conditions:

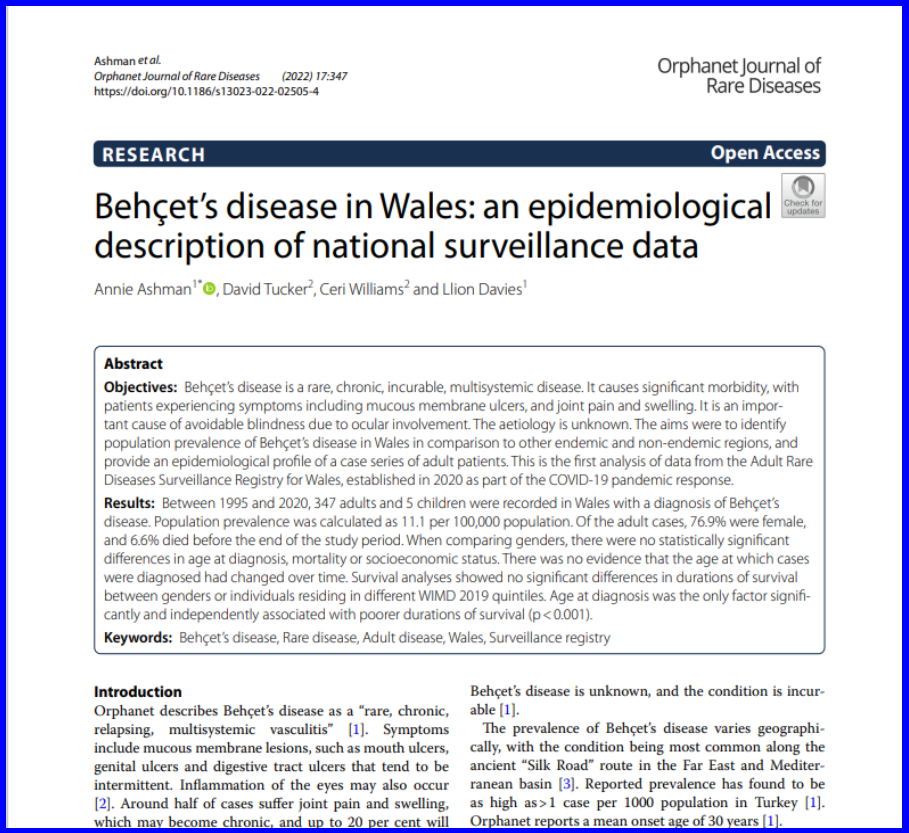
- **Genetic**
- **Immunodeficiency**
- **Chromosomal**
- **Syndromes**
- **Congenital neoplasms**
- **Musculoskeletal**
- **Other rare diseases**

For details on denominators, important caveats and other useful links, click on the **Further Information** tab above.
A guide on how to search for a specific condition/disease can be found by clicking the **Search tips** tab.

- **Cystic Fibrosis**
  - **n=310, 3.96 per 10,000 live births**
- **Angelman**
  - **n=24, 0.3 per 10,000 total births**

Adult Rare Diseases

- First publication
- RDIG
- Sarcoid
- 4 Nations Research
- Co-production



Ashman *et al.*
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Orphanet Journal of Rare Diseases

RESEARCH Open Access

Behçet's disease in Wales: an epidemiological description of national surveillance data

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Abstract

Objectives: Behçet's disease is a rare, chronic, incurable, multisystemic disease. It causes significant morbidity, with patients experiencing symptoms including mucous membrane ulcers, and joint pain and swelling. It is an important cause of avoidable blindness due to ocular involvement. The aetiology is unknown. The aims were to identify population prevalence of Behçet's disease in Wales in comparison to other endemic and non-endemic regions, and provide an epidemiological profile of a case series of adult patients. This is the first analysis of data from the Adult Rare Diseases Surveillance Registry for Wales, established in 2020 as part of the COVID-19 pandemic response.

Results: Between 1995 and 2020, 347 adults and 5 children were recorded in Wales with a diagnosis of Behçet's disease. Population prevalence was calculated as 11.1 per 100,000 population. Of the adult cases, 76.9% were female, and 6.6% died before the end of the study period. When comparing genders, there were no statistically significant differences in age at diagnosis, mortality or socioeconomic status. There was no evidence that the age at which cases were diagnosed had changed over time. Survival analyses showed no significant differences in durations of survival between genders or individuals residing in different WIMD 2019 quintiles. Age at diagnosis was the only factor significantly and independently associated with poorer durations of survival ($p < 0.001$).

Keywords: Behçet's disease, Rare disease, Adult disease, Wales, Surveillance registry

Introduction

Orphanet describes Behçet's disease as a "rare, chronic, relapsing, multisystemic vasculitis" [1]. Symptoms include mucous membrane lesions, such as mouth ulcers, genital ulcers and digestive tract ulcers that tend to be intermittent. Inflammation of the eyes may also occur [2]. Around half of cases suffer joint pain and swelling, which may become chronic, and up to 20 per cent will

Behçet's disease is unknown, and the condition is incurable [1].

The prevalence of Behçet's disease varies geographically, with the condition being most common along the ancient "Silk Road" route in the Far East and Mediterranean basin [3]. Reported prevalence has found to be as high as >1 case per 1000 population in Turkey [1]. Orphanet reports a mean onset age of 30 years [1].

Reference: M*A*S*H, TV show developed by Larry Gelbart, Produced by 20th Century Fox Television, 1972-1983