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## Background

In Scotland, management of most adults with DM1 is delivered through specialist clinics run by four Regional Genetic Services (Fig 1.). All patients ever seen in Genetics clinics or tested in Scottish laboratories are entered on our database and common data are collected when patients attend clinic.

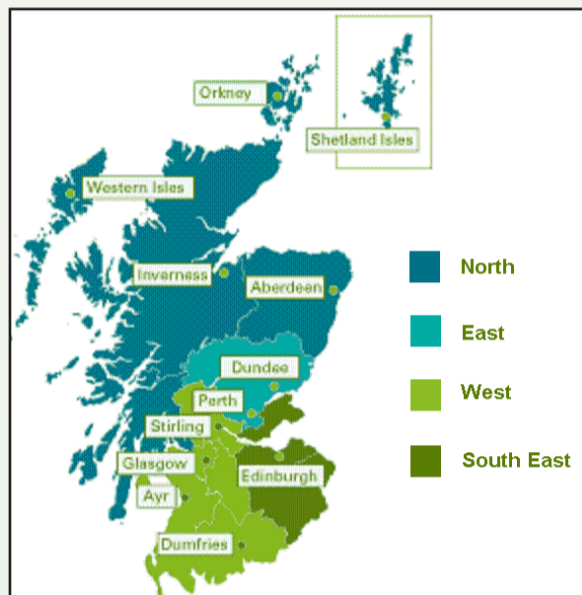


Fig 1. Catchment areas of the four Regional Genetic Services in Scotland

## Objectives

To analyse the clinical features and management of DM1 adults seen at our specialist clinics, observing any regional variation.

## Methods

Data analysis was performed in June 2013 using the electronic patient management databases held by each centre. There were 630 patients known to be living in Scotland with DM1, of which 456 had been seen at least once in a specialist clinic. The data from their most recent attendance were analysed.

## Results and Conclusions

Region	Total number known affected (adults and children)	Prevalence	Total patients seen in dedicated clinic	Congenital Onset	Mean age (years)
West	302	1 in 8609	211 (70%)	26 (12%)	46.1
South East	150	1 in 8000	106 (71%)	6 (6%)	45.5
North	132	1 in 6060	108 (82%)	6 (6%)	45.6
East	46	1 in 10,869	31 (67%)	2 (6%)	46.9
All Scotland	630	1 in 8095	456 (72%)	40 (6%)	46.0

Fig 2. Demographics

Total prevalence of DM1 in Scotland was found to be 1 in 8,095, in line with the widely quoted prevalence of around 1 in 8,000.<sup>1</sup> Mean age was similar in all regions.

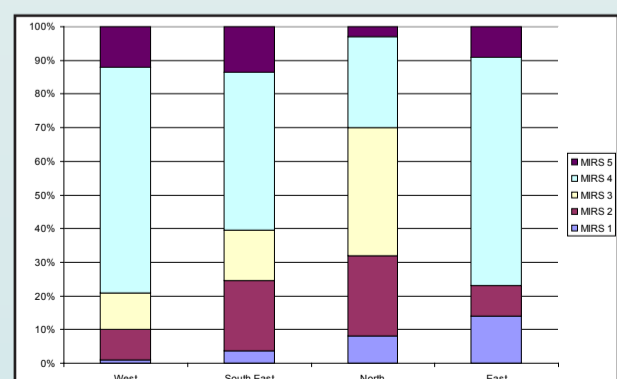


Fig 3. Muscle Impairment Rating Scale (MIRS) scores by region

## Results and Conclusions cont.

A marked variation in MIRS score was observed, with patients in the North in particular scoring lower than other cohorts. This may reflect a true clinical difference, particularly as there is a history of Ophthalmologists in the North pro-actively referring all patients with early-onset cataracts for DM1 testing potentially leading to early diagnosis of an otherwise asymptomatic subgroup. While all clinicians involved in MIRS scoring had undergone 8 hours training by two consultant neurologists, our results could equally reflect inter-user variability in MIRS scoring.

Region	Total with pacemaker	Total with implantable cardiac defibrillator	Under Cardiology management
West	24 (11%)	0	151 (72%)
South East	3 (3%)	4 (4%)	30 (28%)
North	11 (10%)	0	21 (19%)
East	4 (13%)	0	6 (19%)

Fig 4. Cardiology

The proportion of individuals with a permanent pacemaker (PPM) or implantable defibrillator (ICD) was in the region of 10% in all areas, consistent with insertion rates observed in comparable populations in USA.<sup>2</sup> This is despite quite different models for cardiac screening and follow-up in the various regions.

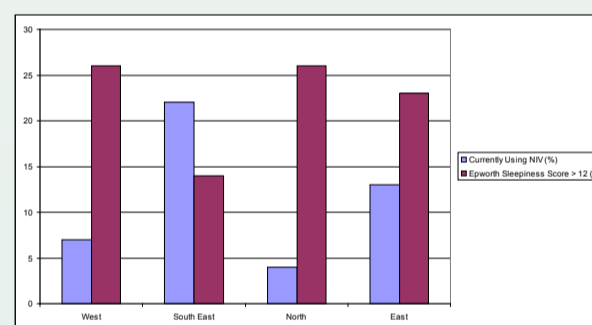


Fig 5: Percentage of patients currently using non-invasive ventilation by region, and comparison with Epworth Sleepiness Score

The proportion of patients actively using NIV in the South East of Scotland is greater than any other region. Epworth Sleepiness Scores >12 were least common in the same cohort. The South East clinic is the only Scottish clinic where patients are routinely seen by specialist ventilation nurses for symptom assessment, spirometry and sniff inspiratory pressures. In addition, it may be relevant that three quarters of the ventilated patients in the South East are managed by a unique nurse-led domiciliary ventilation service. Further in-depth observations are required.

Region	In employment	At college
Glasgow	54 (26%)	11 (5%)
Edinburgh	32 (30%)	6 (6%)
Aberdeen	34 (31%)	4 (4%)
Dundee	12 (39%)	1 (3%)

Fig 6. Social Data

Employment rates for individuals affected by DM1 are around half of the Scottish average for 16-64 year olds at 32% vs 72.2%.<sup>3</sup>

## Conclusion

A national patient management database allows the comparison of both clinical and management data between regional cohorts, highlighting variations and thus potential targets for service improvement.

## References

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