Prevalence of late-onset dystonia
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Uses of epidemiology
- To describe patterns of health and disease in populations (time, place, person) (who gets the disease?)
- To identify causes of disease (risk factors) (why do they get it?)
- To measure the need for health services, their use and their effects. (effectiveness) (what can we do about it?)

Prevalence studies (1)
- Prevalence – the number of people with disease of interest at a specific time point (“point”) or over a time period (“period”) divided by the number of people at risk of developing the disease.
- It is ascertained from cross-sectional studies and is a proportion not a rate (i.e. Incidence-number of new cases divided by person years) Provide data by age, sex, ethnicity, SES etc
- Potential clues to aetiology (genetic or environment, clusters) and enable rationale service planning
**Prevalence studies (2)**

- Cases – standardised case definition; from tertiary, secondary, primary care or door-to-door studies; diagnostic expertise
- Population studies least biased but very time consuming and expensive with imprecise estimates (wide 95% CIs) due to small numbers
- Issues around misdiagnosis and under-diagnosis if based on medical ascertainment
- For comparison of studies need age-specific or age standardised rates

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**Incident cases**

- New diagnosis
- Immigration of ill people

**Prevalent cases**

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Under certain conditions:

- Prevalence
- Incidence x average disease duration
- Cure
- Death
- Emigration of ill people

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**A prevalence study of primary dystonia in eight European countries**

The Epidemiological Study of Dystonia in Europe (ESDE) Collaborative Group

<table>
<thead>
<tr>
<th>Age groups (years)</th>
<th>Cervical</th>
<th>Blepharospasm</th>
<th>Writer’s cramp</th>
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</thead>
<tbody>
<tr>
<td></td>
<td>Male</td>
<td>Female</td>
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</tr>
<tr>
<td>20-49</td>
<td>63.7%</td>
<td>30.3%</td>
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<tr>
<td>50-59</td>
<td>32.8%</td>
<td>67.2%</td>
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<td>60-69</td>
<td>30.3%</td>
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<td>70-89</td>
<td>25.5%</td>
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Age and sex specific rates per million: Cervical 57, Blepharospasm 36 and writer’s cramp 14 per million

Type A centres higher rates than type C centres

Epidemiology of primary dystonia

Giovanni Defazio, Giovanni Abbiruzzese, Paolo Livrea, and Alfredo Berardelli

Table 2: Prevalence rates per million screened for focal onset primary dystonia

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<th>Country</th>
<th>Methodology</th>
<th>Age at onset</th>
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Data are from the International of Neurology Society's database on primary dystonia. The incidence is for all patients treated.

Lancet Neurol 2004; 3: 673–78

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Can we make sense of data from these 12 studies?

- population size (median 195.65, mean 898.606, range 707 to 5,792,937)
- Number of cases (median 30, mean 116, range 2 to 677)
- Rate per million (median 117, mean 757, weighted mean 131, range 30 to 7,320)
- Context: 58% Europe, 8% N. America, 25% Far East, 8% Middle East
- Type of study: 25% population, 42% Neuro plus, 25% Neuro, 8% Linkage
Can we explain outliers?

- Muller et al reported prevalence of 7,320 per million from South Tyrol. Is there something unusual about this population?
- Door-to-door survey with only 6 cases identified (2 previously diagnosed)
- Proportion based on population >50 years (around 41% of total population)
- Crude rate (all ages) = 3,480 per million
- Crude rate (all ages, prior diagnosis=1160 per million, 95% CI 140, 4180 per million)
North England Study

- Next highest rate from study by Butler et al at 521 per 100,000. (error in Defazio paper)
- Not truly cross-sectional due to continuous monitoring of small population from 1993 to identify all cases (hence repeated surveys which will uncover missed cases from past – c.f. MS prevalence studies in Scotland)
- These rates may be closer to the truth – assuming no diagnostic error

Minimum incidence of primary cervical dystonia in a multiethnic health care population

Marras et al Neurology 2007

Results: We identified 66 incident cases of cervical dystonia from 8.2 million person-years of observation. The minimum estimate of the incidence of cervical dystonia in the population is 0.80 per 100,000 person-years. Ethnicity-specific incidence rates were calculated for individuals over age 30. Incidence was higher in white individuals (3.23 per 100,000 person-years) than in persons of other races (0.15 per 100,000 person-years, p = 0.002). The minimum estimated incidence was 2.5 times higher in women than in men (1.14 vs 0.45 per 100,000 person-years, p = 0.0003). There was no significant difference in reported symptom duration prior to diagnosis between women and men (1.4 vs 1.5 years).
Conclusions

- We know relatively little about the epidemiology of focal dystonias other than age and gender differences
- Very few high quality international studies and data on ethnicity and SES still limited and under-diagnosis still an issue
- Potential for genetic and environmental interaction remains to be determined
- Do we need more prevalence studies?
- The ability to undertake large-scale record-linkage studies allows greater potential for aetiological studies