



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?)

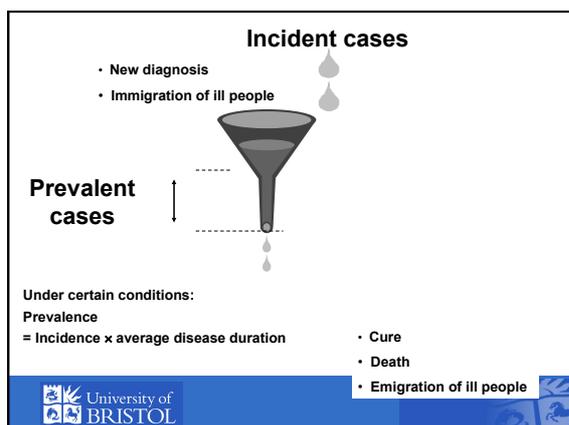


Prevalence studies (1)

- 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



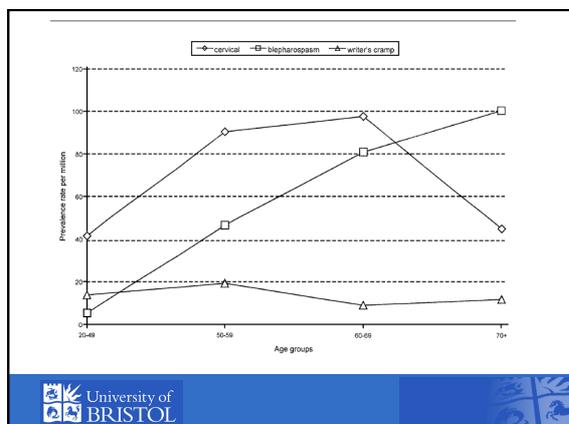
A prevalence study of primary dystonia in eight European countries

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

Age groups (years)	Cervical		Blepharospasm		Writer's cramp	
	Male	Female	Male	Female	Male	Female
20-49	(63) 39.9	(73) 42.8	(7) 4.4	(10) 5.9	(20) 12.7	(25) 14.6
50-59	(32) 77.5	(48) 101.8	(13) 31.5	(28) 59.4	(12) 29.1	(5) 10.6
60-69	(33) 90.9	(43) 103.9	(21) 57.9	(42) 101.5	(5) 13.8	(2) 4.8
70-95	(9) 25.5	(29) 59.0	(22) 62.3	(63) 128.2	(8) 22.7	(2) 4.1

Age and sex specific rates per million: Cervical 57, Blepharospasm 36 and writer's cramp 14 per million

University of BRISTOL Type A centres higher rates than type C centres
 J Neurol (2000) 247:787-792



Epidemiology of primary dystonia

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

Table 2. Prevalence rates per million (crude estimates) from studies on late-onset primary dystonia

Study	Nation, years of study	Study design	Population sample	Age	Distribution of dystonia	Cases	Prevalence estimate (95% CI)
Li et al ¹¹	China, 1983	Door to door	63 195	All ages	Focal	2	30 (4-110)
Nutt et al ¹²	USA, 1952-80	Record-linkage system	406 976	All ages	Focal	17	209 (172-479)
Nakashima et al ¹³	Japan, 1988-93	Neurology-service based	245 000	All ages	Focal	15	61 (34-101)
Khandi et al ¹⁴	Egypt, 1988-90	Door to door	42 000	All ages	Focal	4	100 (26-243)
ESDE ¹⁵	Eight European countries, 1996-07	Neurology-service based	5 792 937	>20 years	Focal	677	117 (108-126)
Defazio et al ¹⁶	Italy, 1987-99	Neurology-service and non-neurology-service based	67 606	All ages	Focal/segmental	9	133 (61-253)*
Castalon-Konkewitz et al ¹⁷	Germany, 1996-07	Neurology-service based	1 807 000	All ages	Focal/segmental	182	101 (84-119)
Müller et al ¹⁸	Italy, 2000	Population sample	707	>50 years	Focal/segmental	6	7320 (3190-15640)
Matsumoto et al ¹⁹	Japan, 2000	Neurology-service and non-neurology-service based	1 459 130	All ages	Focal/segmental	146	101 (84-118)
Dung Le et al ²⁰	Norway, 1999-2002	Neurology-service and non-neurology-service based	508 726	All ages	Focal/segmental	129	254 (212-301)
Pekmezovic et al ²¹	Serbia, 2001	Neurology-service and non-neurology-service based	1 602 226	All ages	Focal/segmental	165	136 (116-159)
Butler et al ²²	England, 1993-2002	Record-linkage system	101 766	All ages	Focal/segmental	43	430 (306-569)

Data were taken from the original report or calculated according to available information. *The estimates refer to blepharospasm alone.



Lancet Neurol 2004; 3: 673-78

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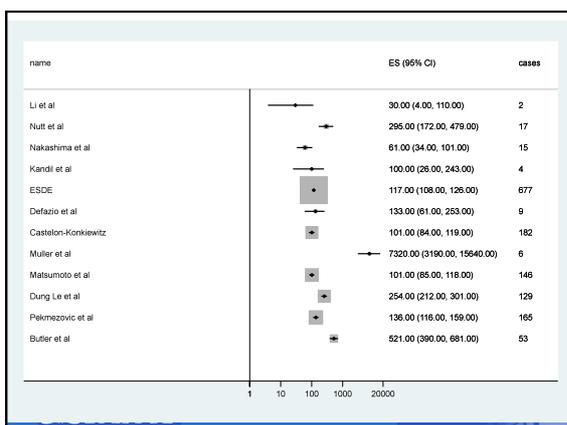
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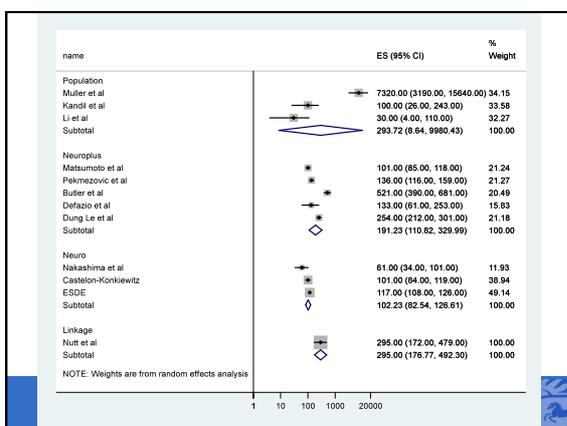


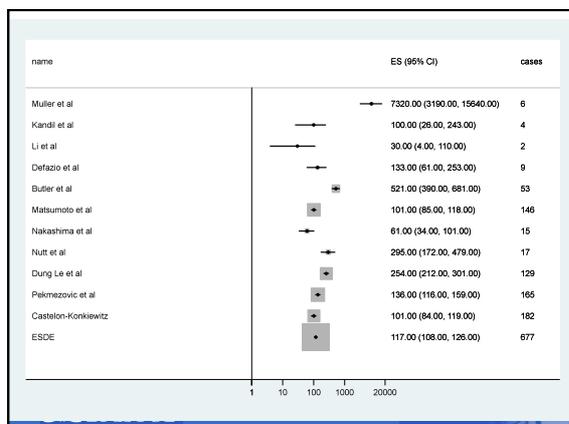
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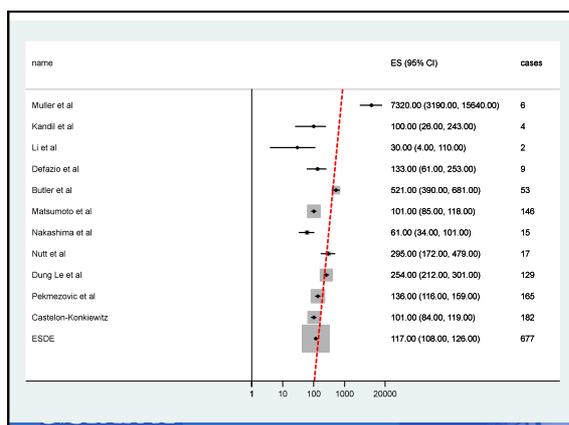
Can we make sense of data from these 12 studies?

- 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









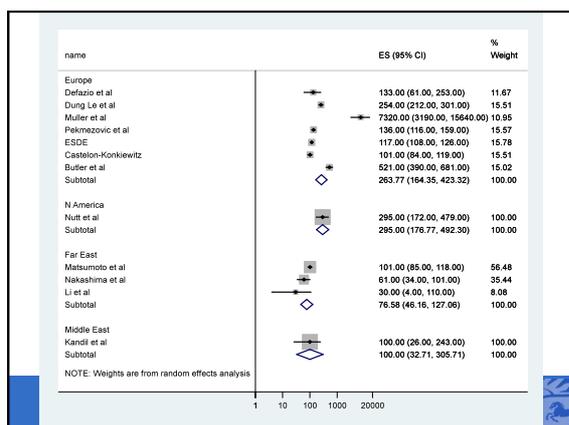
Can we explain outliers?

- 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 this 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 (1.23 per 100,000 person-years) than in persons of other races (0.15 per 100,000 person-years, $p < 0.0001$). The minimum estimated incidence was 2.5 times higher in women than in men (1.14 vs 0.45 per 100,000 person-years, $p = 0.0005$). The average age at diagnosis was higher in women (56 years) than in men (45 years, $p = 0.0004$). There was no significant difference in reported symptom duration prior to diagnosis between women and men (3.9 vs 5.3 years).