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NHS Lothian

Snoring, sleepiness and behavioural correlates in Scottish adults with Down's Syndrome

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Overview

- Background
  - Sleep-disordered breathing
  - Down's Syndrome
- Current study
  - Methods
  - Results
  - Conclusions
- What next?
- Questions

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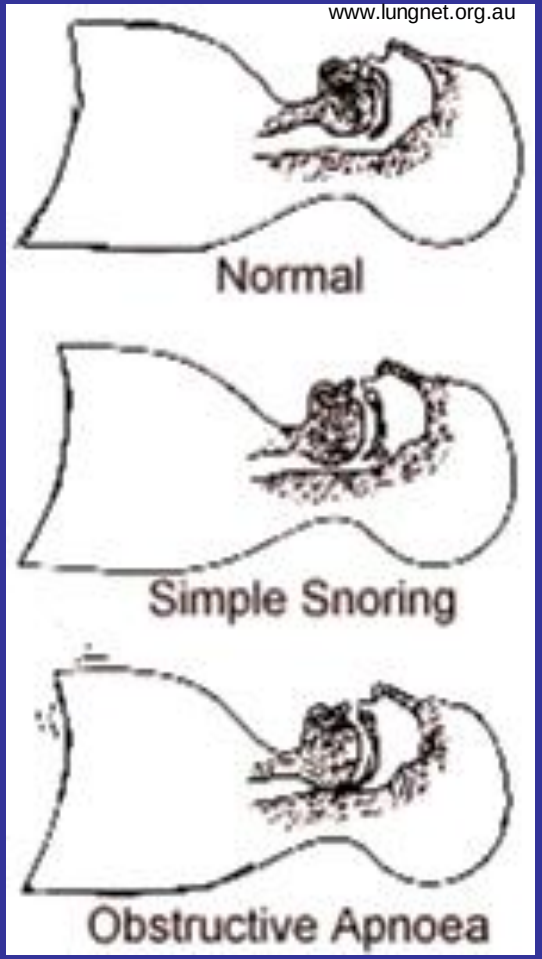
Sleep-disordered breathing

- Repeated pauses in breathing during sleep
- Repetitive cycle of airway obstruction, followed by resumption of breathing
- Affects around 20% of general adult population

Mechanism

- Muscle relaxation during sleep
- Partial (hypopnoea) or complete (apnoea) blocking of airway
- Partial blocking causes vibration → snoring
- Exacerbated by
  - Supine position / gravity
  - Extra fat around neck
  - REM sleep
  - Alcohol
  - Anatomical features



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Terminology

- Obstructive sleep apnoea (OSA)
  - Significant number of pauses in breathing, but not associated with significant sleepiness or other symptoms
- Obstructive sleep apnoea/hypopnoea syndrome (OSAHS)
  - OSA causing significant daytime sleepiness or other symptoms
- Simple snoring
  - Snoring in the absence of OSA

Sleep-disordered breathing

- Prevalence in adult population
  - 2-4% OSAHS
  - 20% SDB
- More prevalent in males than females (2:1)
- Affects all ages but most common in middle age

OSAHS

Daytime symptoms

- Excessive daytime sleepiness (EDS)

↓

- Cognitive impairment
- Personality changes
- Mood disturbances
- Reduced quality of life

Nocturnal symptoms

- Snoring
- Witnessed apnoeas
- Choking / gasping
- Frequent awakenings
- Restlessness / movements
- Nocturia
- Dry mouth (in morning)

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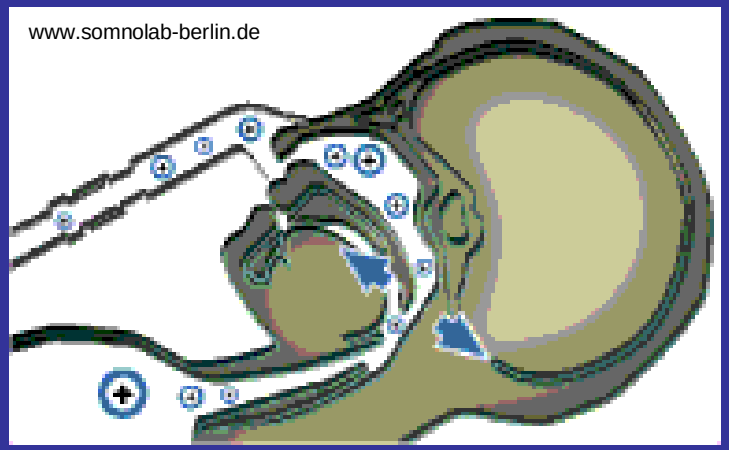
Continuous Positive Airway Pressure

CPAP

- First choice therapy for moderate & severe OSAHS



Down's Syndrome & SDB

- Genetic disorder – chromosome 21
  - 1 in 1000 live births in Scotland *Carrothers, 1994*
- Children and adults with DS are predisposed to SDB
- Related to physiology and anatomy
  - Obesity
  - Facial structure – small midface
  - Thick neck
  - Narrow palate
  - Adenotonsillar hypertrophy
  - Macroglossia
  - Generally reduced muscle tone
  - Increased mucosal secretions

Down's Syndrome & SDB

- Untreated SDB causes cognitive impairment in general population
  - OSAHS severity linked with poorer cognitive performance in adults *Engleman et al, 2000*
  - 10 point IQ deficit in children with SDB v. controls *Kohler et al, 2009*
- Likely that untreated SDB will worsen cognitive impairment already present in some people with DS

Down's Syndrome & SDB

- In general population, sleepiness can manifest as
  - learning difficulties *Curcio, Ferrara & De Gennaro, 2006*
  - behavioural/emotional disturbances *O'Brien, 2011*
- From empirical observations, we hypothesise that the same may be true in the adult DS population

Down's Syndrome & SDB

- Prevalence of SDB in children with DS ~55%

  De Miguel-Diez, 2003

- Prevalence unknown in adults
  - Broken sleep in 7% of DS adults

    Boyle et al, 2010
  - AHI >15 in 88% & ESS >10 in 63% of adults with DS (n=16)

    Trois et al, 2009
  - AHI >10 in 83% of adults with DS (n=6)

    Resta et al, 2003

Down's Syndrome & SDB

- National Institute for Clinical Excellence (NICE) guidelines for recommend CPAP as first-line treatment for adults with OSA
  - Based on evidence from general middle-aged population
  - Did not include any subpopulations, eg. younger people, older people, people with ID…

- Very few studies of CPAP in children with DS, and none in adults
  - Improved ESS, behaviour and QOL scores in subset of 10 children with neurodevelopmental disability (6 with DS) *Marcus et al, 2012*

Down's Syndrome & SDB

There is a need for good quality research studies in adults with DS to address gaps in the current evidence base…

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Current study

Current study

Controlled prospective trial of the effectiveness of continuous positive airway pressure therapy in adults with Down's Syndrome

ISRCTN55685305

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Current study

Research questions:

- Does CPAP use in DS adults with OSAHS/SDB improve sleepiness and quality of life more effectively than lifestyle measures alone?
- What are the potential barriers to implementing CPAP effectively in DS adults with OSAHS/SDB?

Current study: prevalence

- Adults with Down's Syndrome
  - age ≥16 years
- Scotland
  - Population ~ 5.2million

National Records of Scotland, 2011

- 2-part study
  - Prevalence → questionnaire

    +/- home sleep study
  - Treatment → randomised trial of CPAP

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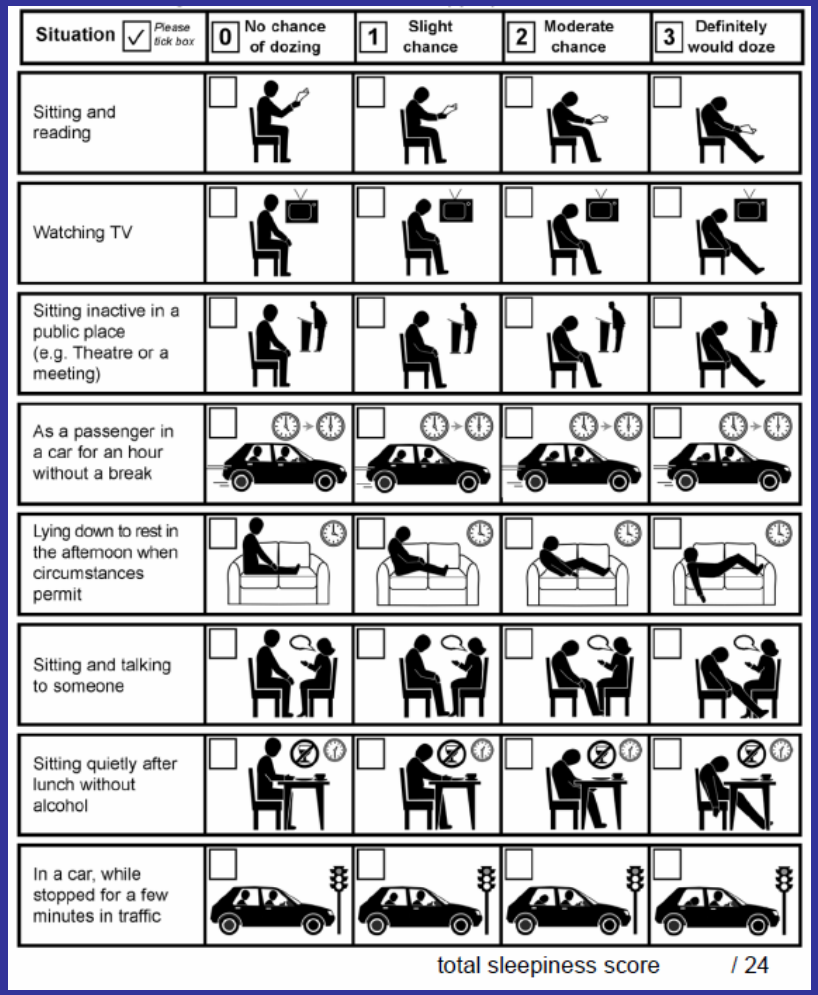
Current study: prevalence

- Easy-read questionnaire
- 2 sections
  - Person with DS
  - Relative/carer

Current study: prevalence

- Person with DS:
  - Medical history
  - Sleep history
  - pESS *Ghiassi et al, 2011*



Situation	Please tick box ✓	0 No chance of dozing	1 Slight chance	2 Moderate chance	3 Definitely would doze
Sitting and reading	☐	☐	☐	☐	☐
Watching TV	☐	☐	☐	☐	☐
Sitting inactive in a public place (e.g. Theatre or a meeting)	☐	☐	☐	☐	☐
As a passenger in a car for an hour without a break	☐	☐	☐	☐	☐
Lying down to rest in the afternoon when circumstances permit	☐	☐	☐	☐	☐
Sitting and talking to someone	☐	☐	☐	☐	☐
Sitting quietly after lunch without alcohol	☐	☐	☐	☐	☐
In a car, while stopped for a few minutes in traffic	☐	☐	☐	☐	☐

total sleepiness score / 24

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Current study: prevalence

- Carer/relative:
  - DBC-A
    - Disruptive
    - Anxiety/antisocial
    - Depressive

Mohr, Einfeld & Tonge, 2004

0 = not true as far as you know
1 = somewhat or sometimes true
2 = often or very true

Office Use Only	Please Circle	
1 a c	0 1 2	Appears depressed, downcast or unhappy.
2 a	0 1 2	Abusive. Swears at others.
3 a	0 1 2	Becomes over-excited.
4 a	0 1 2	Cries easily for no reason, or over small upsets.
5 c	0 1 2	Has become confused or forgetful.
6 c	0 1 2	Has become more withdrawn.
7 b	0 1 2	Has nightmares, night terrors or walks in sleep.
8 a	0 1 2	Has temper tantrums, e.g. stamps feet, slams doors.
9 b	0 1 2	Hides things.
10 a	0 1 2	Impatient.
11 b	0 1 2	Inappropriate sexual activity with another.
12 a	0 1 2	Irritable.
13 a	0 1 2	Jealous.
14 b	0 1 2	Lights fires.
15 c	0 1 2	Loss of appetite.
16 c	0 1 2	Loss of enjoyment or interest in usual activities.
17 c	0 1 2	Loss of self-care skills.
18 b	0 1 2	Makes gloomy statements.
19 b	0 1 2	Masturbates, or exposes self, in public.
20 c	0 1 2	Mood changes rapidly for no apparent reason.
21 c	0 1 2	Moves slowly, underactive, does little, e.g. only sits and watches others.
22 c	0 1 2	Not communicating as much as usual.
23 a	0 1 2	Overly attention-seeking.
24 b	0 1 2	Panics. Sweats, flushes, trembles.
25 b*	0 1 2	Poor sense of danger.
26 a	0 1 2	Refuses to go to college, activity centre or workplace.
27 b	0 1 2	Steals.
28 a	0 1 2	Stubborn, disobedient or uncooperative.
29 a	0 1 2	Tense, anxious, worried.
30 a	0 1 2	Throws or breaks objects.
31 a	0 1 2	Tries to manipulate or provoke others.
32 a	0 1 2	Upset and distressed over small changes in routine or environment.
33 a	0 1 2	Very bossy.
34 a	0 1 2	Whines or complains a lot.

Results

Current study: Results so far

Questionnaire study

- Questionnaires sent 660
- Questionnaire responses 299 (50%)
- Questionnaires valid for analysis 244 (37%)

- OSA diagnosed 15
- Treated 10 (excluded from further analysis)
  - Surgery 3
  - Current CPAP 7

Analysis date: 19 July 2012

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Current study: Results so far

- Gender — 139 males; 105 females
- Age — 32±11 years
- BMI — 29.6±7.4 kg/m^2
- pESS — 7±5
- Snoring — 74% ever
  36% often / frequent
- Apnoeas — 24% ever
  (breathing pauses) — 12% often / frequent

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Current study: Results so far

	n	Total	Male	Female	p
		Mean±SD or median (IQR)			
Age (years)	244	32±11	31±11	32±11	0.62
Body mass index (kg/m²)	201	29.6±7.4	28.1±5.4	31.6±9.0	**0.002**
Pictorial Epworth Sleepiness Score	241	5 (3-10)	6 (3-11)	4 (2-8)	**0.01**
DBC-A Disruptive subscale score	240	5 (2-9)	4 (2-9)	6 (2-9)	0.32
DBC-A Anxiety/Antisocial subscale score	240	0 (0-1)	0 (-1-1)	0 (0-1)	0.25
DBC-A Depressive subscale score	240	2 (0-5)	2 (0-5)	1 (0-5)	0.40

Table 1: Characteristics of questionnaire responders by gender

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Current study: Results so far

Males v. females:

- Females significantly heavier
- But males significantly more sleepy
- No other significant gender differences

Current study: Results so far

	Non-snorers	Snorers	p	No apnoeas	Apnoeas	p
	Mean±SD or median (IQR)					
Gender	21 m; 17 f	107 m; 74 f	0.35	49 m; 39 f	34 m; 25 f	0.96
Age (years)	34±12	30±10	0.05	32±10	28±10	**0.005**
Body mass index (kg/m^2)	27.8±6.4	30.1±7.7	0.11	29.9±7.8	29.6±8.3	0.86
Pictorial Epworth Sleepiness Score	3 (2-4)	6 (3-11)	**<0.001**	4 (2-6)	8 (4-13)	**<0.001**
DBC-A Disruptive subscale score	4 (2-9)	5 (2-9)	0.83	4 (1-7)	7 (3-10)	**0.02**
DBC-A Anxiety/Antisocial subscale score	0 (-1-0)	0 (0-1)	0.88	0 (-1-1)	0 (-1-1)	0.97
DBC-A Depressive subscale score	2 (0-4)	1 (0-5)	0.34	0 (1-4)	3 (1-6)	**0.02**

Table 2: Characteristics of questionnaire responders by incidence of snoring and witnessed breathing pauses

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Current study: Results so far

Snorers v. non-snorers:

- Trend towards being younger
- Significantly more sleepy

- No significant difference in BMI
- No significant difference in behaviour

Current study: Results so far

Witnessed apnoeas v. no witnessed apnoeas:

- Significantly younger
- Significantly more sleepy
- Score significantly higher on measures of
  - Disruptive behaviour
  - Depressive behaviour
- No significant difference in BMI

Current study: Results so far

- All three behaviour subscales were significantly, but weakly, correlated with pESS
  - Disruptive r = 0.16 p = 0.01
  - Anxiety/antisocial r = 0.15 p = 0.03
  - Depressive r = 0.33 p < 0.001

Current study: Results so far

- All three behaviour subscales were also significantly, but weakly, correlated with snoring frequency
  - Disruptive $r = 0.18$ $p = 0.005$
  - Anxiety/antisocial $r = 0.14$ $p = 0.04$
  - Depressive $r = 0.26$ $p < 0.0001$

Current study: Results so far

- Being a snorer was significantly associated with
  - Higher anxiety/antisocial behaviour score
    - p = 0.03 OR 1.5 (CI 95% 1.0-2.2)
  - Higher pESS
    - p = 0.001 OR 1.3 (CI 95% 1.0-1.4)
  - Being younger
    - p = 0.02 OR 1.0 (CI 95% 0.9-1.0)

Current study: Results so far

- Reported breathing pauses were significantly associated with higher scores on
  - Disruptive behaviour subscale
    - p = 0.04 OR 0.2 (CI 95% 0.1-4.3)
  - Depressive behaviour subscale
    - p = 0.008 OR 0.3 (CI 95% 0.5-3.0)

Early conclusions

Early conclusions

- First large population survey of SDB in adults with DS
- Females less sleepy than males, despite being heavier
- pESS is a useful measure of sleepiness in adults with DS, and is significantly higher in snorers and those reporting apnoeas

Early conclusions

- Those reporting apnoeas score higher on measures of disruptive and depressive behaviour
- Together, reported breathing pauses and snoring are significantly associated with higher scores with all three behavioural subscales
- This supports our hypothesis that sleep-disordered breathing can manifest as behavioural problems in adults with Down's Syndrome

What next?

Study ongoing:

- Questionnaires sent out across rest of UK
  - England
  - Wales
  - Northern Ireland
- Continue recruiting
  - Questionnaires
  - Treatment

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Thank you – any questions?

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