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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www.dsscotland.org.uk
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 $\rightarrow$ 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)
Continuous Positive Airway Pressure

CPAP

- First choice therapy for moderate & severe OSAHS

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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

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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...
Current study
Current study

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

ISRCTN55685305
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 $\geq 16$ years

- Scotland
  - Population $\sim 5.2$ million
  
  *National Records of Scotland, 2011*

- 2-part study
  - Prevalence $\rightarrow$ questionnaire
    $+/ -$ home sleep study
  - Treatment $\rightarrow$ randomised trial of CPAP
Current study: prevalence

- Easy-read questionnaire
- 2 sections
  - Person with DS
  - Relative/carer
Current study: prevalence

- Person with DS:
  - Medical history
  - Sleep history
  - pESS  \textit{Ghiassi et al, 2011}

<table>
<thead>
<tr>
<th>Situation</th>
<th>Situation</th>
<th>Situation</th>
<th>Situation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sitting and reading</td>
<td>Watching TV</td>
<td>Sitting inactive in a public place (e.g. Theatre or a meeting)</td>
<td>As a passenger in a car for an hour without a break</td>
</tr>
<tr>
<td>Lying down to rest in the afternoon when circumstances permit</td>
<td>Sitting and talking to someone</td>
<td>Sitting quietly after lunch without alcohol</td>
<td>In a car, while stopped for a few minutes in traffic</td>
</tr>
</tbody>
</table>

\textit{total sleepiness score} / 24
Current study: prevalence

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

Mohr, Einfeld & Tonge, 2004
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/m²
- pESS: 7±5
- Snoring: 74% ever, 36% often / frequent
- Apnoeas: 24% ever, 12% often / frequent

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### Current study: Results so far

<table>
<thead>
<tr>
<th></th>
<th>n</th>
<th>Total</th>
<th>Male</th>
<th>Female</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Age (years)</strong></td>
<td>244</td>
<td>32±11</td>
<td>31±11</td>
<td>32±11</td>
<td>0.62</td>
</tr>
<tr>
<td><strong>Body mass index (kg/m²)</strong></td>
<td>201</td>
<td>29.6±7.4</td>
<td>28.1±5.4</td>
<td>31.6±9.0</td>
<td>0.002</td>
</tr>
<tr>
<td><strong>Pictorial Epworth Sleepiness Score</strong></td>
<td>241</td>
<td>5 (3-10)</td>
<td>6 (3-11)</td>
<td>4 (2-8)</td>
<td>0.01</td>
</tr>
<tr>
<td><strong>DBC-A Disruptive subscale score</strong></td>
<td>240</td>
<td>5 (2-9)</td>
<td>4 (2-9)</td>
<td>6 (2-9)</td>
<td>0.32</td>
</tr>
<tr>
<td><strong>DBC-A Anxiety/Antisocial subscale score</strong></td>
<td>240</td>
<td>0 (0-1)</td>
<td>0 (-1-1)</td>
<td>0 (0-1)</td>
<td>0.25</td>
</tr>
<tr>
<td><strong>DBC-A Depressive subscale score</strong></td>
<td>240</td>
<td>2 (0-5)</td>
<td>2 (0-5)</td>
<td>1 (0-5)</td>
<td>0.40</td>
</tr>
</tbody>
</table>

*Table 1: Characteristics of questionnaire responders by gender*
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

<table>
<thead>
<tr>
<th></th>
<th>Non-snorers</th>
<th>Snorers</th>
<th>p</th>
<th>No apnoeas</th>
<th>Apnoeas</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gender</td>
<td>21 m; 17 f</td>
<td>107 m; 74 f</td>
<td>0.35</td>
<td>49 m; 39 f</td>
<td>34 m; 25 f</td>
<td>0.96</td>
</tr>
<tr>
<td>Age (years)</td>
<td>34±12</td>
<td>30±10</td>
<td>0.05</td>
<td>32±10</td>
<td>28±10</td>
<td>0.005</td>
</tr>
<tr>
<td>Body mass index (kg/m²)</td>
<td>27.8±6.4</td>
<td>30.1±7.7</td>
<td>0.11</td>
<td>29.9±7.8</td>
<td>29.6±8.3</td>
<td>0.86</td>
</tr>
<tr>
<td>Pictorial Epworth Sleepiness Score</td>
<td>3 (2-4)</td>
<td>6 (3-11)</td>
<td>&lt;0.001</td>
<td>4 (2-6)</td>
<td>8 (4-13)</td>
<td>&lt;0.001</td>
</tr>
<tr>
<td>DBC-A Disruptive subscale score</td>
<td>4 (2-9)</td>
<td>5 (2-9)</td>
<td>0.83</td>
<td>4 (1-7)</td>
<td>7 (3-10)</td>
<td>0.02</td>
</tr>
<tr>
<td>DBC-A Anxiety/Antisocial subscale score</td>
<td>0 (-1-0)</td>
<td>0 (0-1)</td>
<td>0.88</td>
<td>0 (-1-1)</td>
<td>0 (-1-1)</td>
<td>0.97</td>
</tr>
<tr>
<td>DBC-A Depressive subscale score</td>
<td>2 (0-4)</td>
<td>1 (0-5)</td>
<td>0.34</td>
<td>0 (1-4)</td>
<td>3 (1-6)</td>
<td>0.02</td>
</tr>
</tbody>
</table>

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

www.europeanoutdoorgroup.com
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