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Carer reported contemporary health problems in people with severe learning disability and genetic syndromes.

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Abstract

Background

Identifying health problems in people with severe and profound disabilities and genetic syndromes can be problematic. There is limited research to inform healthcare provision and the complex health needs of this population are often unmet. Unmanaged health problems may impact on an individual's quality of life resulting in negative affect.

Specific aims

The aims of this study are to report carer reported contemporary health problems in three genetic syndromes associated with severe and profound disabilities and investigate the association of these problems with affect

Method

Carer reported questionnaire data were collected on health issues and affect in individuals with Angelman, Cornelia de Lange and Cri du Chat Syndromes and a group of individuals with intellectual disability of heterogeneous cause. Health problems were classified using ICD-10 criteria. Negative affect was assessed using the Mood, Interest and Pleasure Questionnaire (MIPQ).

Findings

Half of the total sample reported at least one health problem. The nature of health problems varied across groups with particularly high levels of diseases of the nervous system seen in Angelman Syndrome and diseases of the digestive system in Cornelia de Lange Syndrome. Correlational analysis across the groups revealed a positive association between negative affect and the presence of a health problem.

Discussion

High levels of health problems occur in this population with particular problems associating with particular syndromes. These problems seem to have a negative impact on individual

Carer reported contemporary health problems in genetic syndromes affect, though further research investigating this relationship is needed. Implications for clinical practice are discussed.

Introduction

Research has shown that individuals with intellectual disabilities (ID) are more likely to suffer from health problems than those in the general population. Van Schrojenstein Lantman de Valk *et al.* (2000), for example examined the medical records of 318 individuals with ID and 48 459 individuals without ID during January 1996 and found that the ID group were 2.5 times more likely to have had a health problem diagnosed within that month than the comparison group. Differences were also reported in the prevalence rates of specific health problems between the two groups, with higher rates of neurological, psychological, eye and ear problems seen in the ID group. Furthermore, research that has focussed specifically on comparing the nature of health problems experienced by individuals with and without ID (see Jansen *et al.* 2004 of a review of this literature) has shown that problems such as visual and hearing impairments (Van Schrojenstein Lantman de Valk *et al.*, 1997, Evenhuis *et al.*, 2001) and gastrointestinal disorders (Van Schrojenstein Lantman de Valk *et al.*, 1997, Böhmer *et al.*, 1999, Böhmer *et al.*, 2002) are more prevalent in this population and that individuals with *severe and profound* ID in particular are frequently affected.

However, the identification of health problems in individuals with severe and profound intellectual disabilities presents a significant problem for carers and services. One of the main shortcomings of current health services is that equal access is not always available for these individuals and so their needs are often unmet by healthcare providers (Beange *et al.*, 1995; DOH, 2001). One significant barrier is the current system's reliance on patients themselves to report and seek treatment for health problems. Since individuals with severe and profound ID generally have little or no verbal skills, reporting the presence and nature of a health issue can

Carer reported contemporary health problems in genetic syndromes be problematic (Beange *et al.*, 1995; Lennox & Kerr, 1997). The responsibility is therefore shifted onto carers who may not recognise atypical symptoms such as challenging behaviour that could be indicative of an underlying health problem (for example hyperactivity as a symptom of gastroesophageal reflux in individuals with Cornelia de Lange Syndrome reported by Luzzani *et al.*, 2003). Another barrier is created by the unfamiliarity and lack of confidence felt by many healthcare staff when treating individuals with an intellectual disability (DOH 2001). In addition to the struggle to overcome communication difficulties met by staff and patients, staff may also lack knowledge about the particular health problems associated with the aetiology of an individual's intellectual disability. Consequently, treatable problems can often go unrecognised and untreated (Van Schrojenstein Lantman-De Valk *et al.*, 1997; DOH, 2001; Lennox & Kerr 1997).

Although there are studies detailing the prevalence of particular health problems within the ID population as a whole, little research has focussed on health issues in individuals with severe and profound disabilities. One group of individuals with severe and profound disabilities who are likely to encounter numerous health problems, particularly in older age, is individuals with genetic syndromes (Henderson, 2004). Many studies have noted associations between specific medical conditions and particular genetic syndromes. For example Jackson *et al.* (1993) note a preponderance of gastrointestinal and cardiovascular problems in individuals with Cornelia de Lange Syndrome and Cornish and Bramble (2002) report a significant over representation of congenital scoliosis, gastrointestinal and cardiovascular problems in individuals with Cri du Chat Syndrome. However, research in this area is disparate with few comparisons of health problems made across different syndromes using similar measures and large samples. Carrying out such a comparison will reveal important information about the everyday health problems experienced by individuals with particular genetic syndromes. This in turn would inform

Carer reported contemporary health problems in genetic syndromes healthcare provision and practitioners could be made aware of health issues that are related those syndromes.

Previous studies have used a variety of methods to examine the health problems associated with particular genetic syndromes and each method has its own limitations. Conducting direct clinical assessments of individuals in a particular syndrome group generates detailed information but is time consuming and often results in a small and potentially biased sample. Utilising databases held by general practitioners in order to report the types of problems people consult their general practitioner for help with allows for a larger sample size but only allows comment on individuals who are utilising primary care services. The current study describes carer reported contemporary health problems so that insight can be gained into the health issues that are of immediate concern to carers. These are likely to be those that appear to be having a significant impact on an individual's quality of life particularly if they are not being managed satisfactorily. For this reason it is beneficial to use an open ended question and then to impose a well established classification system such as the ICD-10.

It is widely accepted that health status should be taken into account when considering an individual's wellbeing, one of the factors argued to contribute to an individual's quality of life (Seltzer & Krauss, 2001). It follows that a preponderance of health problems in a given population could have a negative impact on that population's quality of life. Indeed it has been noted that the health related quality of life of individuals with epilepsy is significantly reduced. (Sabaz *et al.*, 2001). It would also seem reasonable to assume that such a reduction in an individual's quality of life could have a negative impact on affect. It is equally likely that the presence of a health problem has a direct impact on affect particularly if the problem is

Carer reported contemporary health problems in genetic syndromes untreated and causing discomfort to the individual. Evidence to support this association comes from studies linking chronic medical illness to major depressive illness (e.g. Katon, 2003).

However identifying negative affect in non-verbal individuals can be problematic as communication difficulties may prevent individuals from being able to self report on emotional state, or for others to interpret their communications accurately. Recent development of measures has helped us to overcome this difficulty by assessing affect by proxy. The Mood, Interest and Pleasure Questionnaire is a new tool which has been developed specifically for this purpose and has good established psychometric properties (Ross and Oliver, 2003). By using this measure it is possible to examine the relationship between carer reported health problems and affect.

Three commonly occurring genetic syndromes associated with severe and profound disabilities are Angelman, Cornelia de Lange and Cri du Chat syndromes. Angelman Syndrome is caused by a deletion on the maternally derived chromosome 15 at 15q11-13 (Kaplan *et al.*, 1987; Knoll *et al.*, 1989) and is characterised by developmental delay, speech impairment, movement disorder and “behavioural uniqueness” e.g. highly excitable, often laughing or smiling and appearing happy (Williams *et al.*, 1995). Estimates of prevalence range from 1 in 10,000 to 1 in 40,000 live births (Buckley *et al.*, 1998; Clayton-Smith, 1993). Seizures are frequently experienced by individuals in this syndrome group (Williams *et al.* 1995).

Cornelia de Lange Syndrome is characterised by congenital physical malformations (e.g. heart, gastrointestinal and limb abnormalities), growth retardation and neurodevelopmental delay (Jackson *et al.*, 1993). The syndrome is caused by a gene mutation on chromosome 5p13 (Krantz *et al.*, 2004; Tonkin *et al.*, 2004). Prevalence is estimated at 1 in 50,000 live births

Carer reported contemporary health problems in genetic syndromes (Beck, 1976; Beck and Fenger, 1985) and commonly reported health problems involve the eye and ear, the heart and the gastrointestinal system with gastroesophagol reflux being a particular cause for concern (Jackson *et al.*, 1993; Luzzani *et al.*, 2003). Cri du Chat Syndrome is also caused by a deletion, located in the region 5p15.1 – 5p15.3 and affects approximately 1 in 50,000 live births (Niebur, 1978). The main clinical features are: a characteristic cat like cry in infancy, dysmorphic facies and profound global intellectual disability (Cornish and Bramble, 2002). Again there are specific health problems associated with the syndrome including: congenital scoliosis, gastrointestinal and cardiovascular problems, upper respiratory tract infections, middle ear fluid and infections and dental problems (Baird *et al.*, 2001; Cornish & Bramble, 2002).

A common feature in each of these three syndromes groups is poor expressive language (Williams *et al.*, 1995; Goodban, 1993; Cornish & Munir, 1998). As discussed earlier this can make it difficult for individuals to communicate their health problems to others. Although particular medical conditions have been described in clinical descriptions of each of these syndromes, there have been no comparisons of carer reported, contemporary health problems across different syndrome groups using a single measure. The aims of the current study are to report carer reported contemporary health problems in three genetic syndromes associated with severe and profound disabilities and investigate the association of these problems with affect.

Method

Recruitment

The current study was carried out as part of a larger research project comparing various aspects of behaviour across several different syndrome groups. The University of Birmingham, School of Psychology Research Ethics Committee reviewed and approved this project. Two methods

Carer reported contemporary health problems in genetic syndromes of recruitment were employed. Participants were recruited by sending questionnaire packs indirectly to members of the Angelman Syndrome Support, Education & Research Trust (n=320), the Cornelia de Lange Syndrome Foundation (U.K. and Ireland) (n=234) and the Cri du Chat Syndrome Support Group (n=180). This allowed members to remain anonymous until they opted in to the study. The questionnaire comprised a number of informant based measures covering different aspects of behaviour and a demographic questionnaire.

Questionnaires were also sent directly to participants from two previous studies on the behavioural characteristics of CdLS. In the first study a number of professionals, including clinical geneticists, paediatricians, consultant psychiatrists and head teachers working in special schools, were contacted and asked if they would pass on information about the study to potential candidates (i.e. carers of individuals with CdLS whom they identified from their caseload). Information about the current study was sent to 142 carers of individuals with CdLS who had been recruited in this way and had indicated that they would be willing to part in future research. In the second study a comparison group with heterogeneous cause of intellectual disability was recruited. Adults in residential homes and social education centres working with CdLS participants were asked to identify a second child in their class, day centre or residential home, who did not have CdLS but could be matched with the CdLS participant on age, gender and level of ability. Of these comparison participants, those who had agreed to be contacted again (n=142) were invited to take part in the current study.

Participants

The return rate across groups was 35.5% (n=361). Of these 361 questionnaires, 24 (6.6%) were discarded for one of the following reasons: no confirmed diagnosis, an additional chromosomal disorder was present or data were incomplete. Consequently 337 individuals took part in the

Carer reported contemporary health problems in genetic syndromes study (Angelman Syndrome (AS) =114, Cri du Chat Syndrome (CDCS) = 60, Cornelia de Lange Syndrome (CdLS) = 108 and comparison group (C) = 55). Participant characteristics are shown in table 1. Data from the Wessex Scale (Kushlick *et al.*, 1973) categories speech (never a word, odd words only, sentence and normal, can talk but doesn't), mobility (walk by himself: not at all, not upstairs, up stairs and elsewhere), continence (wetting/soiling nights, wetting soiling days: frequently, occasionally, never) and self help (feed himself/wash himself/dress himself: not at all, with help, without help) were used to calculate the proportion of verbal, fully mobile, continent and able (a self help score of 8 or 9) participants in each group. Although there was variability across groups on all of these characteristics it can be seen from table 1 that substantial disability and communication problems were present in all groups.

+++Insert Table 1 here+++

Measures

A demographic questionnaire was included to gain background information on age, gender diagnosis, when the diagnosis had been given, and by whom. This information was used to assign each participant to the correct syndrome group and ensure that no additional diagnoses had been given alongside the syndrome.

The open ended health question was “Has the person had any medical / health difficulties in the last six months? If yes, please give details: “

The Tenth Revision of the International Statistical Classification of Diseases and Related Health Problems or ICD-10 (World Health Organization, 1992) was used to classify the health problems reported by caregivers. The ICD-10 comprises 21 categories of health problem, (e.g. diseases of the nervous system, diseases of the genitourinary system, mental and behavioural

Carer reported contemporary health problems in genetic syndromes disorders). Thirteen of these categories were used to code the health problems reported by carers (the other eight categories were discarded as none of the problems listed within them had been reported by our informants).

The Mood, Interest and Pleasure Questionnaire (Ross and Oliver, 2003) is an informant based questionnaire used to assess mood, interest and pleasure in individuals with severe and profound intellectual disabilities. Its twenty five items require the informant to rate operationally defined observable behaviours to give a total score, a mood subscale score and an interest and pleasure subscale score. A shorter version of this measure was developed (MIPQ-S, Ross, Oliver & Arron, unpublished) in which twelve items from the original measure were selected (six from each subscale) on the basis of their item total correlation and ensuring that all the original constructs of mood, interest and pleasure were included. Items 1, 2, 4, 5, 7, 8, 11, 12, 16, 17, 22 and 23 were selected and this new version showed good internal consistency (Cronbach's alpha coefficients: total = .88, mood subscale = .79, interest and pleasure subscale = .87), test-retest (.97) and inter-rater reliability(.85). External validity data is not yet available.

Each item is rated using a five point likert scale to give a total score of between 0 and 48 where 48 is the maximum score indicating positive affect, interest and pleasure. Similarly the two subscale scores range between 0 and 24 with 24 being the highest possible score. In order to develop a cut off score that would indicate negative affect, scores on the MIPQ-S for 560 participants in five syndrome groups and one group of controls were analysed. An arbitrary cut off at the tenth percentile was determined as a total score of 28. In the same way a cut off score of 16 was calculated for the Mood subscale and a cut off score of 10 was calculated for the Interest and Pleasure subscale.

Carer reported contemporary health problems in genetic syndromes

Analysis

Chi square analyses were employed to examine comparability on age, gender, verbal skills, mobility, ability and continence (as measured by the Wessex Scale). A series of Fisher's Exact tests were performed in order to compare the prevalence of each health problem (as categorised by the ICD-10) in each syndrome group against the comparison group (a conservative alpha level of $p < .001$ was set). T-tests were used to compare total and subscale scores on the MIPQ-S in participants with and without a reported health problem (alpha level set at $.05/3 = .017$). In each case Levene's test for equality of variances was significant so adjusted t values were used. Participants who scored below the 10th percentile on total score were identified and assigned to a very low affect group. Similarly those who scored below the 10th percentile on the two subscales were identified and assigned to very low mood and very low interest and pleasure groups. Fisher's exact tests were performed in order to compare the prevalence of health problems in individuals with and without very low affect (alpha level set at $.05/3 = .017$). Odds ratios were also calculated in order to determine effect size. Two tailed tests were used throughout all analyses.

Results

Participant characteristics

Chi square analyses revealed significant differences in age (Kruskal-Wallis test $X^2(3) = 16.9$; $p = .001$) gender ($X^2(3) = 14.1$; $p = 0.003$), ability ($X^2(3) = 32.9$; $p < .001$), verbal skills ($X^2(3) = 52.5$; $p < .001$), and continence ($X^2(3) = 33.5$; $p < .001$), across the four groups. No significant difference was seen in mobility scores ($X^2(3) = 6.9$; $p = .074$). A series of Mann-Whitney U tests were performed comparing age in each syndrome group to that in the comparison group. A significant difference was found for the Angelman group ($Z = 3.81$; $p < .001$) where mean age was lower than that in the comparison group.

Reported Health Problems

Across the total population surveyed, 169 (50.1%) participants reported at least one health problem. The percentage of each type of health problem (as classified by the ICD-10), within and across groups, is shown in table 2. The most commonly reported problems within the total sample were: diseases of the digestive system (58.2%), diseases of the respiratory system (53.5%) and diseases of the nervous system (42.8%). The number of health problems reported varied between groups (AS median=0, CDCS median=0, CdLS median=1, Comparison median=0) with the CdLS group reporting significantly more problems than the other groups (Kruskal-Wallis $X^2(3) = 35.88$; $p < .001$). In order to compare the reported prevalence of particular problems in each syndrome group to that in the comparison group a series of Fisher's Exact tests were performed. Fisher's Exact Test revealed a significantly higher number of diseases of the nervous system reported in the AS group than in the comparison group ($p < .001$). Similarly a higher number of diseases of the digestive system was reported in the CdLS group than in the comparison group ($p < .001$). In summary, results show that half of the total population reported at least one health problem and the type of problem reported varied widely. Further analysis revealed a high prevalence of diseases of the nervous system in individuals with Angelman Syndrome and a high prevalence of diseases of the digestive system in individuals with Cornelia de Lange Syndrome.

+++Insert Table 2 here+++

Health problems and affect

Carer reported contemporary health problems in genetic syndromes. The MIPQ-S total score was used to gain an indication of negative affect. The mean total score for the total population was 36.2 (SD 6.9). This score was significantly lower in individuals who had reported a health problem (mean=35.1) than in those who had not (mean=37.5) ($t(321)=3.20, p=.002$). The mean mood subscale score was also significantly lower in individuals who had reported a health problem (mean=19.3) than in those who had not (mean=20.4) ($t(319)=3.08, p=.002$). Similarly the mean interest & pleasure subscale score was also significantly lower in individuals who had reported a health problem (mean=15.8) than in those who had not (mean=17.1) ($t(326)=2.67, p=.008$).

Using the 10th percentile as a cut off point (see measures) in order to form three groups of participants with negative affect (very low affect, very low mood and very low interest & pleasure), the association between presence of one or more health problems and negative affect was examined. Fisher's Exact Test revealed significant differences in the number of participants reporting a health problem in all three groups when compared to the rest of the sample. More participants in the very low affect group (determined using the total MIPQ-S score) had reported a health problem (73.2%) than in the rest of the sample (47.3%) ($p=.001$). A significant odds ratio of 3.04 was calculated (95% confidence level (1.45 – 6.30)) indicating that individuals with a health problem were approximately three times as likely to be experiencing very low affect. This finding was replicated in the very low mood group (determined using the mood subscale score) where the percentage of participants reporting a problem in the very low mood group (69.6%) was higher than that in the rest of the sample (47.4%) ($p=.004$; odds ratio=2.54 (1.27-5.07)) and in the very low interest & pleasure group (determined using the interest & pleasure subscale score) where again the percentage of participants reporting a health problem in the very low interest & pleasure group was higher (69.8%) than that in the rest of the sample (47.6%) ($p=.005$; odds ratio=2.54 (1.30-4.95)).

Discussion

There are no studies in the current literature that examine contemporary health problems across different genetic syndromes through carer report. By asking the same question across three different syndrome groups, each with a large number of participants, we were able to compare the prevalence of health problems across these different syndromes. In support of past research we identified high levels of diseases of the nervous system reported in the Angelman Syndrome group (Leitner and Smith, 1996; Ruggieri and McShane, 1998) and high levels of diseases of the digestive system reported in the Cornelia de Lange Syndrome group (Luzzani *et al.*, 2003). We also found that half of the total population reported at least one health problem. We conclude that there are both associations between particular categories of health problem and particular genetic syndromes, and an association between health problems in general and this population of individuals with severe and profound disabilities. Similarly few studies have examined the association between prevalence of health problems and very low mood in individuals with genetic syndromes. Using a reliable and valid behavioural measure of low mood we were able to look at the impact that these health problems might have on psychological well-being, in a population of individuals unable to report their own emotional status. Here we found that health problems were reported significantly more often in a population of individuals with very low mood.

Our finding that a large number of different health problems are evident across all syndrome groups adds to the literature describing the phenotypes of these genetic syndromes. The two most striking associations (diseases of the nervous system in Angelman Syndrome and diseases of the digestive system in Cornelia de Lange Syndrome) are in accordance with past findings in papers describing the phenotypes of these two syndromes. For example Leitner and Smith

Carer reported contemporary health problems in genetic syndromes (1996) report a clear history of epilepsy in 19 (79%) out of twenty four patients attending an Angelman Syndrome clinic in Sydney, Australia. In this study questionnaire, history, physical examination and diagnostic test data were collected and a clear history of epilepsy was noted in nineteen patients. Similarly in a questionnaire study examining parents' views of epilepsy in Angelman Syndrome (Ruggieri and McShane, 1998), epilepsy was reported by parents in sixty eight out of seventy eight children (87%). These levels are much higher than the level of diseases of the nervous system reported in our AS group. This may be because we didn't ask carers about specific health problems so ongoing conditions such as epilepsy may not always have been reported.

This may also have been the case in the CdLS group where the rate of diseases of the digestive system (32.4%) is lower than that suggested by other studies. For example, Luzzani *et al.* (2003) examined forty three patients with CdLS and found pathological gastroesophageal reflux in 65% of those patients. A correlation between behavioural symptoms and esophageal damage was also seen. They suggest that medical complications such as gastroesophageal reflux cause serious discomfort to the patient and that if this discomfort cannot be resolved, behavioural problems may ensue. It is worth noting that although there were no other significant associations found between a particular health problem and a particular genetic syndrome that is not to say that other health problems were not present at high levels. Numerous and diverse health problems were reported by participants in all groups, including the comparison group, so the level of health problems in the syndrome groups, whilst not significantly different to that in the comparison group, is still high. Furthermore it is possible that overall levels of health problems may be lower than those reported in similar studies due to the current study's reliance on carer report. This issue is discussed later.

Carer reported contemporary health problems in genetic syndromes

The idea that health problems can impact on an individual's quality of life is explored in this study by assessing affect in participants with and without a carer identified health problem. It is very difficult to evaluate the signs and impact of a health problem on a person's emotional state when that person is unable to communicate. Whilst this is often the case in individuals with severe and profound disabilities, observable behaviour might correlate well with affect. By using the MIPQ-S we were able to assess mood, interest and pleasure through reports of observable behaviours such as positive/distressed vocalisations, smiling/crying and flat expression. Our finding that individuals with a health problem scored significantly lower on both subscales and on the total scale of the MIPQ-S indicates that health problems may indeed have a negative effect on an individual's affect. By isolating participants with very low affect (as arbitrarily determined by a cut off point at the 10th percentile) we were able to demonstrate a strong association between the presence of a health problem and very low affect.

A limitation of the current study is that an open-ended question was used to gain information about health problems. A full medical examination was not carried out and neither was a close-ended checklist used to identify whether or not each type of health problem was present. This is because the emphasis of this particular study was to examine health problems, as reported by carers in a particular time window, rather than to conduct a more general survey. The advantage of this approach is that health problems that may be ongoing and are of particular concern to carers are examined rather than health problems that have occurred but may have been resolved or adequately managed. However it is important to note that the prevalence rates reported in this study will not reflect the true prevalence rates of health problems in this population as we have not carried out full medical examinations of our participants and therefore cannot report health problems that parents' are unaware of.

Carer reported contemporary health problems in genetic syndromes
Another consideration is the possibility of a selection bias as a result of our recruiting participants through parent support groups. It is possible that parents have joined a support group because of their children's complex health needs and that is why more problems have been identified in these populations. However the prevalence of health problems we have reported in particular syndromes is actually lower than those reported in the literature on those syndromes suggesting that this bias is not evident.

Another possible criticism of this study is that significant differences exist in the age, gender and abilities of the groups being compared. However, the purpose of the study was to describe health problems reported in different syndrome groups. As we are describing different syndromes it is inevitable that these differences will occur. The difference in age seen between the Angelman group and the comparison group is unlikely to be a potential confound.

A final consideration is the possibility that carers are more likely to be alerted to health problems by participants' low affect, although it is also possible that low affect might mask health problems so that they are actually underreported in this group. Further research on this association between very low affect and the presence of health problems could clarify whether or not we have identified a group at risk. If individuals with very low affect are more at risk then specific interventions, such as annual health checks, could be targeted at this group.

It is clear that health problems occur frequently in individuals with genetic syndromes and severe and profound disabilities with 50% of our sample reporting at least one problem. Furthermore particular health problems occur more frequently in particular syndromes. As the majority of individuals in these groups have poor verbal skills and may be unable to report

Carer reported contemporary health problems in genetic syndromes these health problems, it is important that healthcare staff be made aware of these risk factors.

A review of the 1996 report “Building Expectations: opportunities and services for adults with a learning disability” published by The Mental Health Foundation (2000), made several recommendations concerning ways in which health services could be made more accessible to individuals with an intellectual disability. It highlights concerns that primary care groups may not have enough detailed knowledge of health issues in individuals with intellectual disabilities and therefore may not be able to commission services that will meet the complex health needs of this population. This lack of knowledge is therefore perpetuated so that frontline staff are often left feeling unable to meet the needs of these individuals. In a recent survey of GP’s educational needs in intellectual disability health carried out by Phillips *et al.*, (2004) several areas of health care were identified as being poor, for example preventative and primary health care and health care for complex medical problems. GP’s reported feeling that they had not received adequate training in these areas and the authors argue that this information should be used to inform GP education.

Another recommendation of the report is that GP’s keep a register of patients with intellectual disabilities and offer annual health checks to this vulnerable group. This approach is described in a study by Webb and Rogers (1999) in which the IHC New Zealand Inc., an organisation which provides services to people with intellectual disability, implemented a health screening procedure for all of its residents in an attempt to address the difficulties encountered in providing good quality health care through general practice. Preliminary results showed that that 73% of those screened required follow up interventions, usually health promotion activities, though a number of life saving procedures were also conducted such as a mastectomy to treat a previously undetected breast cancer. The lack of health promotional activities accessed by people with intellectual disabilities is also noted by Leeder and Dominello (2005)

Carer reported contemporary health problems in genetic syndromes who point out that not only do health promotion activities often pass people with ID by they also often do not reach the poorer sections of society and as people with ID are more likely to fall into this category they are likely to be doubly disadvantaged.

And so the importance of focussing on this vulnerable group and finding alternative ways of identifying health problems and promoting good health becomes apparent. Particularly when the knock on effects of poor health are considered, such as low mood and social consequences such as the negative impact medical conditions have on children's engagement in educational activities (Zijlstra and Vlaskamp, 2005).

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Legend for figures

Table 1 Participant characteristics across three genetic syndrome groups and a comparison group.

Table 2 Frequency of report for each type of health problem across three genetic syndrome groups and a comparison group.

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Syndrome Group	n	Mean age in years (range)	% female	% verbal*	% fully mobile*	% able*	% continent*
Comparison	55	18.5 (6 – 38)	36.4	43.6	37.0	27.3	51.9
Angelman Syndrome	114	12.5 (2 – 45)	44.7	0	43.2	0	11.5
Cri du Chat Syndrome	60	17.2 (1 – 44)	65.0	18.6	52.7	6.7	22.4
Cornelia de Lange Syndrome	108	16.6 (1 – 40)	59.3	27.2	56.2	13.0	33.3

* Data derived from the Wessex Scale categories: speech, mobility, continence and self help (Kushlick, Blunden and Cox, 1973)

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ICD-10 classification	Control (n = 55)	AS (n = 114)	CDGS (n = 60)	CdLS (n = 108)	Total (n = 337)
	%	%	%	%	%
Infectious & parasitic	0	0.9	0	6.5	2.4
Blood & blood forming organs/immune mechanism	1.8	0	0	0.9	0.6
Endocrine, nutritional & metabolic	1.8	0	1.7	2.8	1.5
Mental & behavioural	0	0	1.7	5.6	2.1
Nervous system	9.1	23.7 *	1.7	8.3	12.5
Eye & adnexa	1.8	0	0	5.6	2.1
Ear & mastoid process	1.8	0.9	1.7	7.4	3.3
Circulatory system	0	0	0	0.9	0.3
Respiratory system	9.1	9.6	20.0	14.8	13.0
Digestive system	7.3	3.5	15.0	32.4 *	15.4
Skin & subcutaneous tissue	0	2.6	0	5.6	2.7
Musculoskeletal system & connective tissue	0	3.5	5.0	3.7	3.3
Genitourinary system	0	0.9	5.0	5.6	3.0

* significant difference (p< .001) between syndrome group and control group.