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The expression and assessment of emotions and internal states in individuals with severe or profound intellectual disabilities

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ABSTRACT

The expression of emotions and internal states by individuals with severe or profound intellectual disabilities is a comparatively under-researched area. Comprehensive or standardized methods of assessing or understanding the emotions and internal states within this population, whose ability to communicate is significantly compromised, do not exist. The literature base will be discussed and compared to that applicable to the general population. Methods of assessing broader internal states, notably depression, anxiety, and pain within severe or profound intellectual disabilities are also addressed. Finally, this review will examine methods of assessing internal states within genetic syndromes, including hunger, social anxiety, and happiness within Prader–Willi, Fragile-X and Angelman syndrome. This will allow for identification of robust methodologies used in assessing the expression of these internal states, some of which may be useful when considering how to assess emotions within individuals with intellectual disabilities.

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1. Introduction

The focus of this review is the expression of emotion and internal states by individuals with severe or profound intellectual disabilities and the corresponding methods of assessment. A review of this area is important in order to evaluate the current state of empirical literature and highlight the need for further research. It will be argued that researchers and clinicians should be proficient at directly assessing and interpreting the expression of internal states of individuals within this population whose ability to communicate is, by definition, significantly compromised. Expertise in this area would reduce reliance on informant opinion when making decisions with regard to diverse and fundamental issues such as physical and mental health, choice and quality of life (Ross & Oliver, 2003).

This paper opens with a broad overview of the expression of internal states and emotions; their development and role within infancy. The review will then focus on expression of emotions within individuals with severe or profound intellectual disabilities. A systematic review is then described with critique of methodology. Physical and behavioral (e.g., body posture) indicators of internal states will be discussed and evaluated and the clinical implications considered. Due to the paucity of the literature on emotions, the review will then be extended to consider methods of assessing internal states such as anxiety, depression, and pain within this population.

Finally, the review will highlight the range of methodologies and measures used to assess internal states within individuals with genetic disorders[1] associated with intellectual disabilities. This is a valuable and rich literature base as many genetic disorders have well-documented atypical internal states, including excessive hunger and social anxiety, that have been successfully researched using a variety of methods. Each of these methods could further inform the literature regarding emotions and emotional expressions within individuals with severe or profound intellectual disabilities.

While there is a substantial body of literature discussing the capacity of individuals with intellectual disabilities to recognize or label other people's internal states, this will not be covered within this paper. For reviews see Owen, Browning, and Jones (2001) and Zaja and Rojahn (2008).

1.1. Basic internal states and emotions

There has been debate within the literature as to what constitutes the “basic” or “fundamental” human emotions. The widely accepted basic human emotions are happiness, fear, anger and sadness, with disgust and surprise also accepted by most (Turner, 2000) but there is little consensus concerning interest, anticipation, guilt, and shame. Using mostly anecdotal evidence, Darwin (1872) suggested that since the expression of basic emotions, particularly facial expressions, are similar in humans around the world, they must have a hereditary basis. He proposed that emotions serve a function and therefore should be seen as adaptive and, arguably, essential to the survival and reproduction of species. Turner (2000) has expanded upon this work, suggesting that behavioral and facial expressions are critical to group harmony as they effectively communicate emotions, feelings, and intent. This emotion-signaling hypothesis is attracting growing interest and support (e.g., Consedine, Magai, & Bonanno, 2002).

1.2. Facial expressions of emotions

Facial expressions of emotions play a key role in early development as they allow for communication between the pre-verbal infant and their caregiver (Spangler, Emlinger, Meinhardt, & Hamm, 2001). In order to quantify and use the information afforded by facial expressions shown by infants for the purpose of research, reliable, valid, and efficient coding systems must be used (Cohn, Zlochower, Lien, & Kanade, 1999). Two systems, both of which require extensive training procedures, code observable changes in the face: the System for Identifying Affect Expression (AFFEX; Izard, Dougherty, & Hembree, 1983; formerly known as the Maximally Descriptive Facial Movement Coding System (MAX; Izard, 1979)) and the Facial Expression Scoring Manual (FESM; Izard, 1971;1977) and the Baby Facial Action Coding System (Baby-FACS; Oster, 2000; based upon the Facial Action Coding System (FACS; Ekman & Friesen, 1978)). Using various methods, good face and current validity are reported (Ekman, Friesen, & Ancoli, 1980).

Using these coding systems, researchers have begun to identify the development of facial expressions in neonates and babies. Although crying is usually the first demonstration of an emotion (Oster, Hegley, & Nagel, 1992), the associated facial expression is usually coded as undifferentiated distress (Oster et al., 1992). The earliest facial expressions of emotions seen in newborns (aged 3–10 h) are in relation to taste; both positive in response to sucrose and negative in response to quinine solutions (Steiner, Glaser, Hawilo, & Berridge, 2001).

Increasing complexity of emotion expression emerges throughout the first two years of life. Using the FESM, basic emotions can be elicited in infants ranging in age from one to ten months (Hiatt, Campos, & Emde, 1979; Izard, Huebner, Risser, McGinnes, & Dougherty, 1980). These emotions and expressions become functionally organized in relation to the infant and their social context (e.g., Bennett, Bendersky, & Lewis, 2005), resulting in responses that are more differentiated and specific (e.g., Witherington, Campos, & Hertenstein, 2001). This coincides with the development of several important cognitive concepts, including understanding others' intentions (Izard, Hembree, Dougherty, & Spizzirri, 1983), a concept of self, emotion regulation (Stifter & Spinrad, 2002) and elaboration of an internal “affective map” (e.g., Gendler-Martin, Witherington, & Edwards, 2008). Infants begin to label their own emotions from 18–20 months of age (Bretherton, Fritz, Zahn-Waxler, & Ridgeway, 1986), a skill that is highly correlated with cognitive abilities (Bennett et al., 2005) and social behavior (e.g., Fine, Izard, Mostow, Trentacosta, & Ackerman, 2003; Izard et al., 2001).

Alongside having a strong communicative function, facial expressions influence the perceived attractiveness of an infant (Power, Hildebrandt, & Fitzgerald, 1982) and consequently affect adults' reactions to them (Hildebrandt & Fitzgerald, 1978). For example, children rated as more physically attractive are treated more favorably (e.g., Adams, 1977). Infants showing positive facial expressions are also rated as “cuter” (Hildebrandt, 1983), although it is

[1] For the purpose of this review, the term genetic disorders refers to intellectual disabilities with an identifiable genetic etiology (as oppose to idiopathic intellectual disabilities).

unknown if or how flexible perceptions of cuteness, and therefore adults' behaviors, are in response to changes in facial expressions.

Diverse methods of emotional expression have been consistently shown to be important during typical development, in terms of both social and cognitive functioning. It is therefore possible that differences or deficits in the expression of emotions will influence these developmental domains. This has been studied in various populations in both child and adulthood, some of which will be discussed below. These studies can broaden understanding of emotional expression and its role in social communication and cognitive development in childhood and beyond.

1.3. Populations within which facial expressions of emotions are altered or impaired

Despite the strong similarities of facial expressions of emotions between and within cultures and, to some extent, species, some populations show impairments. These include individuals with schizophrenia, dementia, Möbius syndrome and those with severe or profound intellectual disabilities. Literature pertaining to the expression of emotions within each of these populations will be reviewed briefly.

1.3.1. Individuals with schizophrenia

Deficits in the expression of emotions are well documented within schizophrenia (e.g., Kring & Neale, 1996; Salem, Kring, & Kerr, 1996; Tremeau et al., 2005) using a range of methodologies (e.g., self-ratings; Schneider, Gur, Gur, & Shtasel, 1995; informant rating scales; Andreasen, 1984a;1984b; and facial coding systems; Berenbaum & Oltmanns, 1992). The significant impairments in facial expression in individuals with schizophrenia are not due to reduced emotional experience; individuals report emotional experiences equal to or possibly greater than individuals without a psychiatric diagnosis (Herbener, Song, Khine, & Sweeney, 2008; Kring, Kerr, Smith, & Neale, 1993), nor are they simply due to side effects of medication (Earnst et al., 1996; Putnam & Kring, 2007). Despite little outwardly observable facial expression, sensitive methods (e.g., electromyography) have identified greater activity in muscles responsible for smiling and frowning in response to positive and negative stimuli respectively (Earnst et al., 1996). There are also recognized deficits in posed emotional expressions; although varying methodologies make it difficult to draw, firm conclusions (see Izard & Dougherty, 1982, for a discussion).

Such difficulties in both spontaneous and posed expressions alongside the dissociation between experience and expression of emotions suggests an underlying physical or neuromotor deficit (Dworkin, Clark, Amador, & Gorman, 1996) rather than a specific deficit in neural or behavioral systems that underpin the experience (Putnam & Kring, 2007). Despite this hypothesis, there has been little research published on effective interventions, medical or psychological, within this area.

1.3.2. Individuals with dementia

Facial expressions of emotions are important communicative tools (Jakobs, Manstead, & Fischer, 1999), particularly in those who are pre-verbal (see Nelson & de Haan, 1997) or for those with impaired speech (Hemsley et al., 2001), such as individuals who develop dementia. As dementia, most notably Alzheimer's disease, progresses, declines are noted in expressive language skills (Faber-Langendoen et al., 1988; Kertesz & Clydesdale, 1994) and consequently the ability to report internal affective states (e.g., pain; see Smith, 2005, for a comprehensive review). Individuals must therefore rely on other methods to communicate.

Facial expressions, both “adaptive” and “inappropriate” (Porter et al., 1996) are enhanced in frequency and intensity in individuals in the early stages of dementia when compared to healthy controls (e.g., Kunz, Scharmann, Hemmeter, Schepelmann, & Lautenbacher, 2007). As the dementia advances, facial expressions gradually decrease (Norberg, Melin, & Asplund, 1986); by end stage dementia it is only possible to identify individual fragments of facial expressions using the FACS. However, even at this late stage, pleasant and unpleasant stimuli evoke differences in heart rate, respiratory rate and skin temperature (Asplund, Norberg, Adolfsson, & Waxman, 1991).

Akin to individuals with schizophrenia, it could be concluded that individuals with end stage dementia may still experience various emotions, but have difficulties expressing them due to neurological change. The task, therefore, for researchers and clinicians is to develop methods of reliably assessing these states within these populations.

1.3.3. Individuals with Möbius syndrome

Möbius syndrome is a rare congenital disorder associated with unilateral or bilateral paralysis of the abducen and facial cranial nerves (Mobius, 1888, cited in Gillberg & Steffenburg, 1989), although involvement from other cranial nerves is common. It is sometimes associated with a mild degree of intellectual disability and co-morbid autism (e.g., Gillberg & Steffenburg, 1989). This facial paralysis, evident from early infancy, means that individuals with this syndrome cannot produce facial expressions of emotions and often have delays or difficulties with speech (Meyerson & Foushee, 1978).

Surprisingly, there are few studies investigating methods of emotional expression within individuals with Möbius syndrome. Sjogreen, Andersson-Norinder, and Jacobsson (2001) observed impairments in facial expressions in all 25 of their participants (aged 2 months to 55 years), ranging from a mild isolated unilateral weakness to profound bilateral paralysis. Szajnberg (1994) visited one child at six, ten, and fourteen months of age and observed interactive play with both parents. He suggests that while the child was developing and adapting alternative ways of affective communication (such as hand gestures and body posture), the lack of facial expressions began to affect social interactions when the child wanted to express more complex feelings. In line with theories of emotional development, impairments in recognizing others' facial expressions are also noted (Calder, Keane, Cole, Campbell, & Young, 2000; Giannini, Tamulonis, Giannini, Loiselle, & Spirtos, 1984).

Cole (2008) discusses the potential impact that difficulties with expression may have on the experience and development of complex emotions. Surgery and biofeedback mechanisms are continually being trialed and reported but with small samples only (e.g., Zuker, Goldberg, & Manktelow, 2000). These areas require further investigation, combining the results of multiple methodologies (e.g., scanning, self-report, observations) in order to reach valid conclusions which may help inform interventions within this, and other populations, for whom emotional expressions are impaired.

1.3.4. Individuals with severe or profound intellectual disabilities

Understanding and assessing emotions of individuals with intellectual disabilities[2] is within its infancy (Clark, Reed, & Sturmey, 1991). There is considerable debate within the literature as to whether individuals with intellectual disabilities express the same range of emotions in the same way as people without intellectual disabilities. This debate has in part arisen from differences based on level of intellectual disability and, importantly, differences based upon the etiology of the intellectual disability. What has not been ascertained is whether there is a difference in the experience or expression of emotions within individuals with intellectual disabilities. This review is therefore particularly pertinent, both from a clinical and a research perspective, for understanding if and how,

[2] The term “intellectual disability” will be used throughout this thesis in preference to a number of other terms (learning disability, mental retardation, mental handicap and mental deficiency) in line with current usage of terminology in recent Department of Health publications.

people without language and accompanying cognitive impairment communicate their internal states. Such knowledge, albeit in its infancy, can form the basis for assessing (and potentially intervening in) subjective experience that is likely to be strongly associated with an individual's quality of life.

We now review the literature base pertaining to the expression of emotions within individuals with severe or profound intellectual disabilities. The quantitative results of a comprehensive literature search are presented and compared to searches undertaken within sub-populations of the general population. The literature is then summarized, describing facial and behavioral expressions of emotions within this population alongside the ability of others to recognize and label such expressions. The expression of emotion within individuals on the autistic spectrum is not discussed specifically within this review but is reviewed by Bormann-Kischkel, Amorosa, and von Benda (1993).

2. Literature search

2.1. Method

Computerized searches were undertaken on Web of Knowledge and Medline. Web of Knowledge searches the titles and abstracts of all journals listed within the Science Citation Index, Social Sciences Citation Index and the Arts and Humanities Citation Index published after 1981. MEDLINE searches the abstracts and titles of over 1800 journals published by Elsevier.

The specified keywords were learning disability/ies intellectual disability/ies, developmental disability/ies, mental retardation, mental handicap and mental deficiency. These were combined with emotion/s, affect, mood, facial expression/s, happy, sad, disgust, surprise, anger and fear. Abstracts were selected if they provided information on expressions of internal states in individuals with severe or profound intellectual disabilities. Once this search was completed, reference lists of the selected papers were also searched. Results of the searches and information retrieved are summarized below.

2.2. Results

The electronic searches on Medline and Web of Knowledge yielded 2544 papers. However, only eighteen of these focused on expression of emotions in individuals with severe or profound intellectual disabilities. Dates of publication ranged from 1987 to 2008. Methodologies employed included behavioral and facial observations, informant based interviews and questionnaires. Details of these eighteen papers are provided in Table 1, which describes, where applicable, the sample size, age range, and level of intellectual disability and summarizes the key findings.

The main topics covered by the 2528 papers that did not focus on the expression of emotions in individuals with severe or profound intellectual disabilities were: the ability of individuals with intellectual disabilities to recognize others' emotions, the emotions of family members or individuals working with people with intellectual disabilities, the expression of emotions in individuals with mild or moderate intellectual disabilities and the adaptation of therapeutic approaches (e.g., cognitive behavioral therapy) for working with individuals with mild or moderate intellectual disabilities.

2.3. Comparison with typically developing population

In order to evaluate the volume of literature pertaining to the expression of emotions within individuals with severe or profound intellectual disabilities, comparison searches were undertaken for two populations. First, neonates (i.e., under one month old) were chosen as they are a population with a discrete body of literature in which the problem of lack of verbal communication is shared. Statistically, they also make up approximately the same proportion of the population (.128% based on data from the 2001 UK census) as individuals with intellectual disabilities (.13%). For this search, the specified keywords of neonate and newborn were combined with the emotion related keywords described above (emotion, affect, mood, facial expression/s, happy, sad, disgust, surprise, anger, and fear).

The second population used for comparison was infants; i.e., under 12 months. This population, although larger in proportion (approximately 1.5% based on data from the 2001 UK census), is similar to individuals with severe or profound intellectual disabilities in that they rely on non-verbal methods to express their emotions. Here, the keyword of infant was combined with the emotion related keywords listed above.

The electronic searches on Medline and Web of Knowledge yielded 12,350 papers for neonate and emotional keywords, and 14,679 papers for infant and emotional keywords. For reasons of practicality, further searches were not undertaken to identify what percentage of these papers specifically addressed the expression of emotions. However, the number of papers identified suggests that the literature regarding the expression of emotions in individuals with intellectual disabilities and, more specifically, severe or profound intellectual disabilities, is comparatively sparse.

3. Expression of emotions and internal states in individuals with severe or profound intellectual disabilities

3.1. Defining severe and profound intellectual disabilities

The DSM-IV (American Psychiatric Association, 1994) and the ICD-10 (World Health Organization, 1992) classify intellectual disability into four categories based upon IQ (or estimates thereof): mild (IQ range 50–70), moderate (IQ range 35–49), severe (IQ range 20–34) and profound (IQ below 20). This review will focus upon literature and research with individuals with severe or profound intellectual disabilities as both language and sensory deficits (the importance of which will become apparent throughout this review) are predominant within this population.

3.2. Methods of identifying emotions within individuals with severe or profound intellectual disabilities

There is now an extensive literature describing the cognitive functioning, adaptive behavior, challenging behavior, and service planning needs of individuals with severe and profound intellectual disabilities. The question must be posed as to why research has not addressed the emotions experienced and expressed within this population, given their perceived importance in other populations (Arthur, 2003; Bailey, Matthews, & Leckie, 1986; Wagner, 1991).

The papers identified in the above literature search (alongside other supporting papers) will now be reviewed. This will allow an overview of the current literature base while highlighting some of the methodological difficulties that may have limited progress within this area.

3.2.1. Facial expression of emotions

Facial expressions are powerful nonverbal channels for conveying emotions (Plesa-Skwerer, Faja, Schofield, Verbalis, & Tager-Flusberg, 2006). Reviewed in Section 1.2, facial expressions of emotions are expressed by typically developing infants at a very early age (Steiner et al., 2001) and can be reliably interpreted by both trained and untrained adults who are familiar or unfamiliar with the infant (Hiatt et al., 1979; Sorce & Emde, 1982).

Deficits in expression of emotions are noted within individuals with idiopathic and genetically determined intellectual disabilities (e.g., Down syndrome; Cicchetti & Sroufe, 1976; Cornelia de Lange

Table 1
Characteristics of the eighteen studies identified.

Authors	Participants (n)	Age range (years)	Level of ID	Summary
Arthur (2003)	N/A	N/A	N/A	Review paper focusing upon the emotional experiences of people with ID. Arthur concludes that "emotional developmental problems and disturbance" are present in a higher number of individuals with ID than originally reported. He suggests that multidisciplinary action should be taken in both assessment and intervention in order to improve quality of life.
Bailey et al. (1986)	N/A	N/A	N/A	Review paper that highlights the paucity of research into emotions in individuals with ID. Authors try to encourage researchers to open up further discussion in this area, taking ideas from our own knowledge as well as carers and individuals with ID. Secondly, they encourage people to begin to work on interventions and activities that help people with ID become more aware of emotions and their own feelings and how they can be expressed through behavior.
Carr et al. (2003)	8	29–48	Mild (1), moderate (3), profound (4)	Observed mood ('good/neutral/bad') across a 30 min observation period. Participants then asked to either complete a task (demand condition) or were allowed to engage in leisure activities for five minutes (no-demand condition). Aim was to see the effect of mood and demands on problem behavior. Only 'bad' mood was predictive of problem behavior.
Charlot et al. (1993)	60	Mean 50	Mild/ moderate (16), severe/ profound (44)	Two groups of 30 participants, those who had a diagnosis of affective disorder, and those who had a diagnosis other than affective disorder. Symptoms of affective disorders were commonly noted, although not always sufficient to meet a diagnosis. Aggression was frequently noted in both groups. Authors suggest the use of modified criteria for depression and mania in individuals with ID and believe these disorders to be underdiagnosed within this population.
Cicchetti and Sroufe (1976)	14	All 18 months	Moderate–profound	Children with Down syndrome laughed at groups of stimulus items in the same order as typically developing children, but delayed. Level of cognitive development was predictive of the level of affective development.
Clark et al. (1991)	75	17–80	Mild (16), moderate (19), severe (20), profound (20)	Study looked at behaviors associated with sadness in individuals with ID. Fifteen item questionnaire on behaviors associated with sadness, items derived from DSM-III criteria and study by Pirodsky et al. (1985). Asked staff if the participant showed each of the behaviors when sad (answered yes/no/not applicable). Two members of staff interviewed per participant. Poor inter-rater agreement for all items, best for verbal abuse (Kappa = .58), physical aggression (.52) and crying (.44). No association between level of ID and behavioral presentation.
Collis, Moss, Jutley, Cornish, and Oliver (2008)	42 (14 CDLS, 14 CDC, and 14 idiopathic ID)	2–15	Moderate–profound	Low levels of emotional expression shown by all three groups during a brief observation sample, especially low levels of facial expressions reflecting negative effect. No difference in duration of positive, negative or flat affect between the groups but ratio of negative to positive affect in CdLS group was significantly higher than two contrast groups.
Davis et al. (2004)	3	31–45	Profound	Compared indices of happiness in three conditions; typical classroom activities, social interaction and social interaction with a preferred stimuli. Confirmed the work of Green and Reid (1996), showing that indices of happiness are related to the environment and available stimuli.
Green and Reid (1996)	6	18–41	Profound	Concluded that individually behaviorally defined markers of happiness (e.g., smiling, hand flapping) are valid as they were more frequent when the individuals with ID were interacting with preferred and non-preferred stimuli. Second study showed that the environment and activities can significantly affect the mood displayed by individuals with ID.
Green and Reid (1999a;1999b)	3	27–66	Profound	Used behavioral indicators of happiness and unhappiness to evaluate the efficacy of an intervention to reduce unhappiness during an exercise routine. The unhappiness reduction intervention was accompanied by decreases in the indices of unhappiness, but only if the intervention was tailored to each individual.
Helm (2000)	N/A	N/A	N/A	Review of measuring happiness in individuals with ID. Highlights importance of individualizing indices of happiness, and where possible, getting first hand descriptions of indices from the individual with ID. Concludes that a combination of quantitative and qualitative methods may be the most productive way of measuring happiness in this population.
Lancioni et al. (2005)	N/A	N/A	N/A	Review paper focusing on research into increasing happiness of individuals with severe/profound ID and multiple disabilities. Identified 24 studies focusing on 6 different methods. Authors combined data in studies and report that increases in indices of happiness were noted in 70–100% of participants, dependent upon the intervention used. Key difficulties with such studies included identifying highly favorable stimuli, small participant numbers, lack of random sampling and low frequency of follow up data.
Liu et al. (2007)	75	19–71	Profound	Assessed the use of the Interact Short Form for evaluating the mood and behavior of people with profound ID. Removed one item and identified three factors (emotional expression, interest towards tasks, behaviors to environment). Good inter-rater (Kappa = .68–.77). Results of modified questionnaire suggest that sample show happy/content behaviors more frequently than less desirable behaviors, such as wandering, restlessness or aggression.
Matson et al. (1991)	69	Not stated	Severe/ profound	Study compared diagnostic criteria on DSM-IV for anxiety to results of DASH-II anxiety scale. DASH-II diagnosed anxiety in 33 individuals, DSM-IV in only 7. Authors conclude that anxiety is particularly difficult to diagnose in this population as only behavioral markers can be used, many of which could be misconstrued. They recommend caution conducting careful observation of the client in different situation alongside a thorough assessment of any confounding factors before considering a diagnosis of anxiety.
Maurer and Newbrough (1987a)	32 adults with LD, and 23 adults without LD	24–62	N/A	Looked at the ability of adults with and without ID to recognize emotions in children with and without ID. Adults with ID were less accurate than adults without ID at recognizing emotional expressions. Facial expressions of children without ID were recognized more accurately than children with ID. This difference was mainly due to difficulties in differentiating between "neutral" and "sad" by adults without ID, and "angry" and "sad" by adults with ID.

(continued on next page)

Table 1 (*continued*)

Authors	Participants (n)	Age range (years)	Level of ID	Summary
Maurer and Newbrough (1987b)	50 (23 inexperienced adults, 21 parents, and 6 teachers)	21–61	N/A	Looked at the ability of adults with varying experience of ID to recognize emotions in children with and without ID (teachers, parents/carers, and adults with no experience). Expressions of emotions were more accurately identifiable in typically developing children than in children with ID. Teachers of children with ID were better at recognizing emotions in children with ID than parents/carers, who were in turn better than inexperienced adults. Children with ID may show more discrete signs of emotion.
Ross and Oliver (2003)	N/A	N/A	N/A	Review paper discussing the assessment of mood in severe/profound ID. Highlight two themes in the literature; firstly that the literature has only recently begun to recognize the presentation of affective disorders in this population. Secondly, the presentation of affective disorders, particularly depression, may be "atypical" in this population, especially in those with profound ID.
Singh et al. (2004)	3	28–44	All profound	Main aim was to see if increasing mindfulness of care staff could increase happiness of participants. Observed baseline measures of happiness (individualized to each participant based upon carer report). Participants showed a higher frequency of behaviors that suggested happiness around staff who had received mindfulness training than those who did not.

syndrome; Johnson, Ekman, Friesen, Nyhan, & Shear, 1976; autism and idiopathic intellectual disabilities; Yirmiya, Kasari, Sigman, & Mundy, 1989). There are two important themes to consider when reviewing this literature: the capacity of individuals with severe or profound intellectual disabilities to communicate emotions through facial expressions; and, perhaps more importantly, the ability of individuals working or living with that individual to interpret and understand such expressions.

3.2.1.1. Facial expressions in individuals with severe or profound intellectual disabilities. There is very little literature investigating the development of expressions of emotions within this population with regard to chronological or mental age. Cicchetti and Sroufe (1976) found a positive association between cognitive and affective development in a longitudinal sample of fourteen infants with Down syndrome. The developmental trajectory of laughing to increasingly complex stimuli was the same as a sample of typically developing infants, but was delayed by a number of months dependent upon degree of intellectual disability. Interestingly, they noticed that those with a greater degree of intellectual disability may only smile under conditions where the more able infants would laugh, suggesting that the expression of emotions may be somewhat more subtle in children within this population. Cicchetti and Sroufe suggest a motor co-ordination element to this delay; research into more able children with Down syndrome has identified specific deficits in eliciting a reliable facial expression of fear (see Sroufe & Wunsch, 1972), although this has not been investigated within individuals with severe or profound intellectual disabilities.

Reliable and valid methods for observing facial expressions may allow clinicians, researchers, and caregivers to develop a better understanding of how an individual with very limited or no expressive language might convey a positive or negative affective state. This could then reduce the reliance on informants when making important or complex decisions (Ross & Oliver, 2003) as even when using structured questionnaire measures, informants' reports differ from those of individuals with intellectual disabilities, especially in relation to more complex internal experiences such as those associated with mental health (Ruddick & Oliver, 2005).

Within the behavioral literature, there are few studies that have observed facial expressions to obtain information about the individual's experience of the intervention (see Ross & Oliver, 2003, for a full review of these studies). Lindauer, DeLeon, and Fisher (1999) observed facial indices of positive and negative affect when evaluating the effects of an enriched environment program. Oliver, Hall, Hales, Murphy, and Watts (1998) noted changes in positive and negative affect before and after an intervention to reduce self-injurious behavior. Facial expressions of emotions have also been used to identify reinforcers or punishers (e.g., Fisher et al., 1992).

Green and Reid (1996) operationally defined facial expressions of happiness and unhappiness in order to evaluate a program aimed at increasing happiness in individuals with profound intellectual disabilities. While happiness is a private event that is not easy to measure directly or manipulate (Kennedy & Souza, 1995), Green and Reid suggest that an individual's public behaviors can give them "clues" as to their private experience. This correlational approach does not allow for definitive conclusions as to whether happiness itself was altered and observed (Green & Reid, 1996) but does provide a starting point for observing such expressions of emotions within this population. Green and Reid increased facial indicators of happiness through their "fun time" program; results which have since been replicated by Davis, Young, Cherry, Dahman, and Rehfeldt (2004) and Ivancic, Barrett, Simonow, and Kimberly (1997).

Lancioni, Singh, O'Reilly, Oliva, and Basila (2005) reviewed 24 studies that focused on interventions designed to increase indices of happiness in individuals with severe or profound intellectual disabilities and multiple disabilities. The majority of the studies reviewed used facial expressions, alongside physical actions (e.g., hand flapping, clapping) and behavior as indices of happiness. Together, these studies suggest that it is possible to increase the indices of happiness within this population. However, the variation in reported efficacy of the six types of intervention reviewed by Lancioni et al. (2005) must serve as a reminder that there is not a "one-size-fits-all" approach for this population, but each intervention should be tailored to that individual and their environment.

This work by Green and Reid (1996,1999a,b) would benefit from further replication, as at present it is limited by small participant numbers and thus potential lack of generalizability. Replicating the interventions with individuals with mild or moderate intellectual disabilities, using self-report or questionnaire methods alongside observations or behavioral expressions of choice, could also provide validation for using such behavioral indicators of happiness.

3.2.1.2. Recognition of facial expressions in individuals with severe or profound intellectual disabilities. The ability to provide reliable, independent indices of emotions becomes pertinent when the literature regarding caregiver recognition and interpretation of facial expressions of emotions in individuals with severe or profound intellectual disabilities is reviewed. Again, a paucity of research within this area limits the extent to which conclusions can be drawn.

The facial expressions of children with severe intellectual disabilities in static photographs are less well recognized than those of typically developing children (Maurer & Newbrough, 1987a), with happy faces being easier to recognize than neutral, sad, and angry faces. Adults inexperienced with individuals with intellectual disabilities were less accurate on all four emotions than parents of children with intellectual disabilities as they tended to label most facial expressions as "neutral" (Maurer & Newbrough, 1987b). Maurer

and Newbrough suggest that parents may be used to interpreting more subtle expressions of emotions that other adults may overlook. Interestingly, there was no difference between parents and inexperienced adults on rating emotions of typically developing children, suggesting that the differences found were not due to general emotion recognition abilities. The authors conclude that children with intellectual disabilities do express their emotions through facial expressions, but experience and careful observation is required in order to interpret and understand them.

It could be argued that interventions within this field should be aimed at improving emotion recognition within families and caregivers as they are best placed to learn and implement new skills (Butterfield & Arthur, 1995). Such caregiver based interventions have previously proven successful (e.g., Singh et al., 2004) but are hampered by high levels of staff turnover and burnout (Hastings, Horne, & Mitchell, 2004); limiting the extent to which such interventions are practical or beneficial.

3.2.2. Behavioral expression of emotions

Developing an understanding of how behavior relates to affect or emotion is important in individuals with few or no verbal skills (Helm, 2000; Ross & Oliver, 2003). Challenging behavior has been associated with communication difficulties within both experimental (e.g., Bott, Farmer, & Rhode, 1997; Quine, 1986) and carer or support staff based questionnaire studies (e.g., Hastings, 1997). This has led to the suggestion that challenging behavior may be analogous to communicating expressions of emotion or emotional distress (e.g., Carr & Durand, 1985; Thurman, 1997).

Lindauer et al. (1999) found a high correlation between indices of negative affect and challenging behavior in their case study of a 23-year-old woman with severe intellectual disabilities and depression. She engaged in self-injurious behavior in 36% of the ten-second intervals in which she displayed negative affect and only 3.2% of the intervals where she did not display negative affect. However, the authors did not conduct a lag analysis in order to establish the sequence of and relationship between these behaviors. Carr, McLaughlin, Giacobbe-Grieco, and Smith (2003) also found a higher likelihood of problem behavior during an intervention if staff rated a participant as being in a "bad mood" earlier in the day. Interestingly, they also note that offering the participant access to stimuli associated with positive moods during the intervention improved mood ratings.

The literature does seem to suggest a relationship between emotions and challenging behavior, although, at present, it is unknown if this is causative or due to an underlying mediating factor, such as physical pain (see an excellent review by Carr & Owen-Deschryver, 2007).

3.3. Methods of assessing internal states in individuals with severe or profound intellectual disabilities

This review has so far established that individuals with severe and profound intellectual disabilities experience a range of emotions and express them through facial expressions and behavior. Such conclusions are mainly based upon observational studies (e.g., Green & Reid, 1996) or independent ratings of videos or photographs (e.g., Maurer & Newbrough, 1987a;1987b). Due to the paucity of research focusing explicitly on the expression of emotions within individuals with severe or profound intellectual disabilities, the remit of the review will be broadened to include other "internal states" including mood, hunger, and affect. Including these broader internal experiences will allow for the discussion of methods of assessment that could potentially be used or adapted to assess emotions within this population.

Due to the degree of intellectual disability, methodologies widely used within the typically developing population are often inappropriate for assessing internal states with individuals with intellectual disabilities. The Interact Short Form — People with Profound Intellectual Disabilities Edition (original version by Baker & Dowling, 1995; adapted by Liu et al., 2007) is an eleven-item observation tool designed to assess internal states before and after interventions or environmental changes. A trained rater observes the individual while performing daily routine sedentary activities (Baker & Dowling, 1995). Internal consistency (.83) and inter-rater reliability (mean Kappa = .72, range .68–.77) are both good although they were based upon a small sample (n = 33 for inter-rater reliability) with very limited variation in mood and behavior.

Comparatively more assessment methods are available for assessing broader internal states, notably depression, anxiety, and pain. Each of these areas will be briefly reviewed below in terms of their presentation (in relation to the typically developing population) and current methods of assessment. A full review of all available assessments within these areas is outside the scope of this review; comprehensive reviews are provided elsewhere for mood (Ross & Oliver, 2003), anxiety (Matson, Smiroldo, Hamilton, & Baglio, 1997) and pain (Symons, Shinde, & Gilles, 2008).

3.3.1. Depression and anxiety

It was only recently accepted that individuals with intellectual disabilities are more susceptible to mental health problems than the typically developing population (e.g., Matson, Gardner, Coe, & Sovner, 1991). At present, estimate rates for mental health problems range from 10 to 60% for individuals with intellectual disabilities (King, State, Shah, Davanzo, & Dykens, 1997). Such high levels of variability are reported due to sampling biases, methodological or diagnostic differences and psychosocial and environmental factors (see Ross & Oliver, 2003). Prevalence rates within children with severe or profound intellectual disabilities are yet to be determined.

Whether depression and anxiety present differently among individuals with severe and profound intellectual disabilities remains a matter of debate. Due to the communication difficulties inherent within this population, obtaining subjective reports of feelings, fears, and thoughts is compromised or impossible (Matson, Smiroldo, et al., 1997). Reliance is therefore placed upon rating scales, informant interviews and potentially, observational techniques (each of which is described in detail elsewhere (e.g., Bramston & Fogarty, 2000; Liu et al., 2007; Ross & Oliver, 2003)) to identify changes in sleep (e.g., Marston, Perry, & Roy, 1997), appetite and low mood (e.g., Charlot, Doucette, & Mezzacappa, 1993).

Matson et al. (1997) highlight some of the problems with differential diagnosis when assessing mental health problems within individuals with severe or profound intellectual disabilities. They highlight numerous symptoms highly suggestive of anxiety within the general population that occur frequently within individuals with severe or profound intellectual disabilities without anxiety. These include sudden motor or vocal responses (which may be part of a stereotypic behavior), trembling or shaking (which may be associated with tardive dyskinesia associated with psychotropic medications; Aman & Singh, 1985;1991), sleep difficulties and exaggerated startle responses (present in many individuals with autism; Schopler, Reichler, & Renner, 1988). Diagnoses should therefore always consider information from informant-based questionnaires alongside a detailed history of behavior and behavioral change.

3.3.2. Pain

Pain is one of the most common experiences of all humans and is, arguably, one of the most difficult to identify (Phan, Edwards, & Robinson, 2005). Akin to the other internal states and emotions, it is a subjective, internal experience that cannot be assessed through direct measurement. The neuroanatomical system required to feel pain is complete by 26 weeks gestation, however, the developmental processes necessary for the mindful experience of pain are yet to occur (see Derbyshire, 2006). This, along with the facial and

behavioral expression of pain, develops throughout childhood, and is shaped by cultural norms (e.g., Schiavenato et al., 2008).

The expression of pain among individuals with severe or profound intellectual disabilities has received considerable levels of attention. Due to degree of intellectual disability, difficulties are present in self-reporting the presence of pain, its intensity (Biersdorff, 1994) and location (Hennequin, Morin, & Feine, 2000). This, coupled with a common misconception that all individuals with intellectual disabilities are insensitive or indifferent to pain (see Foley & McCutcheon, 2004), highlights the need for sensitive assessment methods and integrative translational research within this area (Symons et al., 2008).

There are many instruments that translate the internal experience of pain into quantitative data, including some that have been developed for individuals with severe or profound intellectual disabilities. Defrin, Pick, Peretz, and Carmeli (2004) suggest that behavioral indices of pain can provide a more complete picture of the pain experience in individuals with severe or profound intellectual disabilities. Vocalizations, verbalizations, physical gestures, autonomic responses, general appearance and facial expressions can provide information on the degree of pain and its associated discomfort. Self-injurious behavior is also associated with higher scores on pain measures (Symons, Harper, McGrath, Breau, & Bodfish, 2009).

Bodfish, Harper, Deacon, and Symons (2001) developed the Pain and Discomfort Scale (PADS) to assess pain in individuals with little or no expressive language. The scale relies upon facial expressions and body movements to indicate acute pain and discomfort. Validation studies show promising results for both acute and chronic pain (Bodfish et al., 2001). Phan et al. (2005) conclude that although the instrument is somewhat difficult to use, it is better than using clinical observations alone. Davies and Evans (2001) conclude that the best method of assessing pain in individuals with severe or profound intellectual disabilities is a thorough clinical assessment combined with a familiarity and understanding of both intellectual disabilities and the individual themselves. As pain is likely to be expressed through more than one non-verbal behavior (Davies & Evans, 2001), and may differ between chronic and acute pain (Collignon & Giusiano, 2001), it is important that caregivers and clinicians alike remain flexible with their interpretation and approach.

3.3.3. Summary

The studies reviewed above suggest that individuals with severe and profound intellectual disabilities do experience some extremes of internal states, such as depression, anxiety and pain, but the presentation of these experiences may differ slightly due to the degree of disability. The underlying theme from these three areas (depression, anxiety, and pain) is the importance of recognizing and acknowledging facial and behavioral markers of internal states within this population. Structured behavioral observation schedules with acceptable reliability and validity are required in order to utilize the communicative nature of such expressions of internal state and experience.

3.4. Assessment of internal states within individuals with intellectual disabilities of a genetic etiology

This review has so far identified a lack of literature relating to the expression of emotions in individuals with severe or profound intellectual disabilities. Methods of assessing broader internal states, including depression, anxiety and pain are somewhat better, but remain in their infancy.

If, as is widely accepted within the literature, internal states and emotions have a genetic element (albeit, one that interacts or can be sometimes overridden by the environment), it could be hypothesized that differences in internal states, or their expression, may be present in individuals with genetic, or chromosomal, abnormalities, such as those with intellectual disabilities with genetic etiologies.

Genetic etiologies can be identified for approximately 13–46% of those with a mild intellectual disability and 33.2–70% of individuals with severe or profound intellectual disabilities (see Leonard & Wen, 2002, for an in depth discussion). Many individuals with genetic etiologies will have a profile of behavioral (and physical) features that are highly associated with their genetic cause of intellectual disabilities. Such associations have led to the notion of the "behavioral phenotype", which proposes that the genetic basis of specific intellectual disabilities (syndromes) strongly influences the comparative likelihood of the occurrence of particular behaviors (Dykens, 1995).

The literature pertaining to behavioral phenotypes within individuals with genetic disorders and intellectual disabilities contains descriptions of behaviors that have been researched in depth using multiple methods, which ultimately combine to derive multi-level models. By turning to this literature within this review, we can identify specific behaviors and internal states that have been researched in depth within syndrome-specific populations. This then allows us to identify robust methodologies used in assessing the expression of these internal states, some of which may be useful when considering how to assess emotions within individuals with intellectual disabilities. Three examples of this are discussed in turn; hunger in Prader–Willi syndrome, social anxiety in Fragile-X syndrome, and laughing and smiling in Angelman syndrome.

3.4.1. Assessment of hunger in individuals with Prader–Willi syndrome

Prader–Willi syndrome is a genetic syndrome that arises from the absence of the expression of the paternal genes on chromosome 15q11–q13 (see Boer et al., 2002). Current estimates suggest a prevalence of approximately 1 in 25,000 births and a gender ratio of 1:1 (Whittington et al., 2001). The behavioral and cognitive phenotype has recently been reviewed (see Dimitropoulos et al., 2000) and includes low-average intelligence or a mild to moderate intellectual disability, excessive eating, repetitive and self-injurious behavior, temper outbursts, mood disturbances and underactivity. The physical phenotype includes distinctive facial features, hypotonia at birth, hypogonadism, small hands and feet and small stature (e.g., Cassidy & Driscoll, 2009). After an initial poor suckling relax, hyperphagia onsets in infants between the age of 1 and 6 years (Cassidy, 1997).

It is this hyperphagia that has been the focus of many studies using various methodologies, including informant based questionnaires or interviews, behavioral observations, choice paradigms and PET scanning, all of which will be reviewed briefly below. Together, results have allowed researchers to begin to understand this internal experience in relation to specific environmental, cognitive, and neurological processes and to begin to identify gene-brain-behavior pathways.

Informant based interviews and questionnaires have been designed in order to gain a quantitative measure of hyperphagia using frequency or intensity of behaviors, including food 'sneaking' and theft, eating unpalatable items or taking food out of the bin. Dykens, Maxwell, Pantino, Kossler, and Roof's (2007) thirteen item informant based Hyperphagia Questionnaire considers both behavioral and emotional aspects of hyperphagia, using a scaled response for each answer to allow for comparisons both within and between individuals over time. Russell and Oliver (2003) devised the sixteen item Food Related Problem Questionnaire that focused upon preoccupation with food, impairment of satiety and other food-related "challenging" behavior. With robust psychometric properties (including test-retest and inter-rater reliability), Russell and Oliver recommend the use of this questionnaire for both research and clinical practice to appraise the food-related problems seen within this population. Although most individuals with Prader–Willi have

the language skills to self-report some of their experiences (see Roof et al., 2000), very few studies have used such reports due to issues with reliability and validity (Dykens et al., 2007).

Behavioral observations and choice paradigms allow for the identification of the prevalence and topography of behaviors, often in relation to groups matched on age or ability. Holland, Treasure, Coskeran, and Dallow (1995) gave individuals with Prader–Willi syndrome access to unlimited amounts of food in a laboratory setting and compared their intake with that of individuals without Prader–Willi syndrome or an intellectual disability. Individuals with Prader–Willi syndrome consumed three times more calories and took longer to show behavioral markers of satiety. The internal drive for food is so strong in individuals with Prader–Willi syndrome that they are more likely to choose a large amount of food over a small amount, even if the large amount is less-preferred (Glover, Maltzman, & Williams, 1996) or delayed (Joseph, Egli, Koppekin, & Thompson, 2002) and will even eat contaminated foods or odd food combinations (Dykens, 2000).

Recent neuroimaging work has been used to begin to understand how dysfunction in one or more areas of the brain may lead to specific behaviors. PET scanning studies have identified significant delays in activation (Shapira et al., 2005) or even non-activation (e.g., Hinton et al., 2006) in areas associated with satiety in typically developing controls. When asked to look at pictures of preferred food and imagine eating them, adults with Prader–Willi syndrome did not show any activation in the brain regions associated with food reward or satiety seen in the typically developing control group. Further work using event-related-potentials suggests a difference in food stimulus evaluations between the two main genetic subtypes; those with uniparental disomys focused more on the suitability of the food for eating, while those with paternal deletions focused exclusively on the quantity of food items (Key & Dykens, 2008). This highlights the need to investigate syndrome specific behaviors in relation to genetic subtypes.

Together, the information gathered from all of these methods can be integrated to suggest a link between the underlying genetics of Prader–Willi syndrome, dysfunctional neural satiety systems and the abnormal eating behaviors (Hinton et al., 2006). Such pathways are imperative to begin to understand behaviors within specific populations and consequently, implement effective interventions. In order to advance the literature around emotions and internal states within individuals with intellectual disabilities, such integration is critical.

3.4.2. Assessment of social anxiety in Fragile-X syndrome

Extremes of specific emotions or internal states are reported with several genetic syndromes associated with mild or moderate intellectual disabilities. Mood disorders have been noted within Smith–Magenis syndrome (Bersani, Russo, Limpido, & Marconi, 2007), depression within Down syndrome (e.g., Collacott, Cooper, & McGrother, 1992) and social anxiety in Fragile-X syndrome (e.g., Turk & Cornish, 1998) and Cornelia de Lange syndrome (Richards, Moss, O'Farrell, Kaur, & Oliver, 2009).

Fragile-X syndrome is the most common inherited cause of intellectual disability with a current prevalence of one in 4000–6000 live births (Turner, Webb, Wake, & Robinson, 1996). There are differences in the phenotype between males and females because of the association with the X chromosome (Loesch, Huggins, & Hagerman, 2004), with males showing a stronger phenotype and a greater degree of intellectual disability than females (Alanay et al., 2007). The behavioral phenotype includes language impairment, social anxiety, gaze aversion, repetitive and self-injurious behavior (see Hagerman & Hagerman, 2002).

Again, multiple methods have been used to investigate the internal experience of anxiety within Fragile-X syndrome. Behavioral and observational methodologies have allowed researchers to conclude that individuals with Fragile-X are typically interested in social interactions, but behaviors including aversion to eye contact, self-injury, and avoidance of novel objects or situations (Turk & Cornish, 1998; Turk & Graham, 1997) highlight the difficulties they have within this area. These discomfort or avoidance behaviors can become severe enough to interfere with social functioning and peer relationships, particularly among females with the full mutation. Eye tracking methods have identified that individuals with Fragile-X spent less time looking at eyes and faces as a whole. However, this did not affect their ability to encode information relating to the faces (Holsen, Dalton, Johnstone, & Davidson, 2008).

Aspects of social anxiety within this population have also been investigated in terms of physiology. Galvanic skin responses increase during direct eye contact and decrease during gaze aversion (Belser & Sudhalter, 1995) and levels of cortisol are heightened following socially challenging tasks (Wisbeck et al., 2000). After accounting for a number of other factors, Hessl et al. (2002) identified an association between salivary cortisol concentration and a number of challenging behaviors seen within children with Fragile-X, including withdrawn behavior, social problems and attention deficits (based upon carer report).

The literature is now beginning to map how the genetically derived dysfunction of several specialized neural networks (e.g., Hessl et al., 2002) lead to difficulties in the ability to regulate arousal (e.g., Wisbeck et al., 2000). This in turn may lead to greater reactivity to environmental and social stimuli (Miller et al., 1999), making social interaction uncomfortable and promoting avoidance or escape behaviors (see Cohen, 1995, for a full discussion). Once again, this brief review has demonstrated how integrating the results of multiple methodologies can enhance our understanding and allow us to begin to infer and explore gene-brain-environment interactions of internal experiences within specific genetic syndromes.

However, we cannot rely solely upon behavioral or physiological markers to identify an internal experience. Williams syndrome is a genetically determined intellectual disability associated with a deletion on chromosome 7q11.23. It has a current prevalence of one in 20,000 and most have moderate to mild intellectual disabilities. In contrast to individuals with Fragile-X, individuals with Williams syndrome are eager to engage in social interaction, and will often do so impulsively, even with strangers (Dykens, 2003). Such behaviors persist into late adulthood (Jones et al., 2000) and have been associated with specific genetically controlled neural circuitry (Meyer-Lindenberg et al., 2005).

Although individuals with Williams syndrome have a high propensity to seek social interaction, like individuals with Fragile-X syndrome, they show atypical gaze behavior (albeit different in nature; Riby & Hancock, 2009). Using eye-tracking methodology individuals with Williams syndrome fixed their gaze on the actor's face for a significantly longer period than the mental age matched control group. This result was maintained for moving cartoons, but not for cartoon stills. Abnormal gaze behaviors may therefore reflect discomfort (as in Fragile-X) or intense interest (as in Williams syndrome). This dissociation highlights the need for careful methodology and definitions alongside consideration of etiology when observing or investigating behavior within individuals with intellectual disabilities.

3.4.3. Laughing and smiling behavior in Angelman syndrome

Angelman syndrome is a rare neurodevelopmental disorder first described in 1965 (Angelman, 1965) with a current prevalence estimate of 1 in 40,000 live births (Buckley, Dinno, & Weber, 1998; Clayton-Smith, 1993). It is caused by a disruption of the maternally inherited portion of chromosome 15q 11–13 (Clayton-Smith & Laan, 2003; Knoll, Nicholls, & Lalande, 1989) via four known genetic mechanisms (Jiang, Lev-Lehman, Bressler, Tsai, & Beaudet, 1999; Lossie et al., 2001). The clinical presentation of Angelman syndrome has been described in detail within the literature. The physical

phenotype includes movement or balance disorder (resulting in an ataxic gait), microcephaly, epilepsy and abnormal EEG, hypopigmentation, wide mouth with widely spaced teeth, protruding tongue and high levels of salivation (resulting in drooling).

The behavioral phenotype is reviewed extensively by Horsler and Oliver (2006a). Of note are presence of severe to profound intellectual disability, raised levels of laughing, smiling and happy demeanor, excessive sociability, little or no speech, hyperactivity and aggression in 6–10% (Summers, Allison, Lynch, & Sandler, 1995). Sixty (94%) out of the 64 studies reviewed by Horsler and Oliver identified elevated levels of laughing and smiling behaviors. The question therefore is why this behavior is present, how is it associated with the genetic cause of Angelman syndrome and how can we assess it?

Most genes have the same effect regardless of whether they were inherited from the mother or father. However, parent origin does matter for a small group of genes; a concept referred to as genomic imprinting (Murphy & Jirtle, 2003). The kinship theory (also known as the genomic conflict hypothesis) of genomic imprinting proposes that maternal and paternal alleles can be expressed differently within the offspring in order to favor perpetuation of the maternal or paternal chromosomes (Haig & Wharton, 2003). Paternal alleles ensure survival of the offspring by increasing the cost to the offspring's mother while the maternal alleles promote maternal survival by reducing these costs. Maternal genes on chromosome 15q 11–13 are not fully expressed in individuals with Angelman syndrome; kinship theory, therefore, predicts high rates of behaviors that promote survival of the paternal chromosomes.

It has been hypothesized that some aspects of Angelman syndrome have the aim of increasing probability of gaining maternal resources (e.g., Brown & Consedine, 2004; Haig & Wharton, 2003). Using emotion-signaling theory (which suggests that expressions of emotions communicate internal states of the sender and influence the behavior of the receiver), Brown and Consedine suggest that smiling and laughing behaviors evoke high levels of social contact and, therefore, maternal resources.

It is possible to identify a number of other evolutionary benefits. Positive expressions of internal states are the primary means for social bonding between both children (Malatesta, Culver, Tesman, & Shepard, 1989) and adults (Bachorowski & Owren, 2001). As discussed in Section 1.2, typically developing children who express high levels of positive internal states (such as laughing and smiling) are rated as 'cuter' (Power et al., 1982) and more physically attractive (e.g., Hildebrandt, 1983) and therefore may be treated more favorably (Adams, 1977; Hildebrandt, 1982).

The focus on, and hypotheses about, the function of the laughing and smiling behaviors in children with Angelman syndrome has recently expanded. Early work suggested that these behaviors were neurologically driven, and therefore environmental factors were not influential (e.g., Dooley, Berg, Pakula, & MacGregor, 1981; Williams & Frias, 1982). However, careful experimental manipulation of the environment identified that both the frequency and duration of these behaviors are related to environmental context.

Taking methodologies and procedures from the behavioral literature, Oliver, Demetriades, and Hall (2002) conducted naturalistic observations of three children with Angelman syndrome and found that levels of laughing and smiling are significantly higher during social interaction conditions than in control conditions. Horsler and Oliver (2006b) extended this work, using analogue conditions to experimentally manipulate the environment (notably adult behavior) to observe the effect upon the child's behavior. Through this they identified that higher levels of laughing and smiling behaviors occurred when adult attention, and particularly adult eye contact, was present. Finally, employing sequential lag analyses to naturalistic observation data, Oliver et al. (2007) found that every smile by children with Angelman syndrome ($n = 13$) evoked higher levels of eye contact, smiles and attention from the adult than smiles of children without Angelman syndrome (matched for degree of intellectual disability). This social contact then makes the child smile; possibly as a result of the positive affect and internal reward mechanisms initiated by these interactions (Oliver et al., 2007).

Adams, Horsler, Mount, and Oliver (2010) extended the work of Horsler and Oliver (2006b), investigating the effect of age on smiling and laughing, looking and approach behaviors demonstrated by children with Angelman syndrome. Interestingly, the only child behavior that changed in frequency with age was laughing and smiling which was significantly lower in the older age group than the younger age group, but only in one environmental condition; where the adult talked, touched, and made eye contact. This decline in laughing and smiling behaviors with age is in line with that predicted by the genomic conflict and kinship theory (Brown & Consedine, 2004; Haig & Wharton, 2003). Typically as a child ages and requires less maternal resources, it might be hypothesized that these resource-soliciting behaviors will decrease. It could be hypothesized that as the child with Angelman ages, they find social interaction and eye contact less reinforcing, it therefore evokes less positive affect and subsequently less laughing and smiling behaviors in older children.

Therefore, the smiling and laughing behaviors seen in Angelman syndrome are a highly efficient way of promoting survival of the paternal chromosomes as they are both beneficial to the child (increases investment from mother) and costly to the mother (time lost pursuing the welfare of other children or activities). Robust methods of assessing these behaviors are now being used and, consequently, are providing more information on the exact topography and potential function in relation to genetics and, specifically, genomic imprinting. Further research and integration of genetics, neuroimaging, physiology and behavioral observations may allow us to further understand the gene-brain-behavior pathway. However, due to the inability to self-report, the question remains as to whether these laughing and smiling behaviors reflect an internal experience of positive affect.

3.4.4. Summary

By identifying and reviewing the literature pertaining to the assessment of internal states (hunger, social anxiety, laughing and smiling) within specific genetic syndromes, it has been possible to identify multi-level approaches to assessing associated behaviors. Some of these, such as scanning techniques and eye movement data, are arguably more likely to be used in research than clinical settings. Nonetheless, they have provided essential information for understanding these behaviors from both a neurological and physiological perspective. Other studies, such as those that have identified the distinct pattern of behavior demonstrated by individuals with Fragile-X upon meeting strangers (which highlight the internal experience of social anxiety), questionnaires which focus upon hunger related behavior in Prader–Willi syndrome, and those that identified the association between social contact and higher levels of smiling and laughing behaviors in Angelman syndrome, used methods that have excellent potential to be used clinically. A common thread of all of the studies reviewed within this section is that they use carefully designed measures with excellent reliability and validity, which focus upon a narrow range of internal experience rather than attempting to assess a broad range of behaviors relating to multiple internal experiences.

4. Conclusions

This review has demonstrated the importance of observing and understanding behavioral and facial expressions of internal states in individuals with severe or profound intellectual disabilities. These expressions provide valuable insights into the internal experiences of individuals who cannot directly communicate their thoughts and feelings. The paucity of literature pertaining to the emotions of individuals with severe or profound intellectual disabilities is of concern. Only by expanding the remit of this review to encompass broader internal states could methods of assessment be identified and evaluated.

Examination of the literature regarding the expression of internal states in individuals with severe or profound intellectual disabilities illustrated several points. First, that research within this area is limited. Second, that there may be deficits in both the sending and receiving of facial expressions of internal states within this population. Individuals with severe or profound intellectual disabilities may exhibit more subtle facial expressions of internal states, which adults are poor at interpreting if they do not have experience of caring for, or communicating with, this population. Such difficulties were also noted within the extremes of internal states, notably depression, anxiety and pain. Although symptoms may be slightly atypical it is important to remember that they may reflect the same level of internal experience (and arguably discomfort) and, therefore, should be responded to appropriately.

By reviewing the syndrome specific literature, it has become apparent that there are numerous measures and methods that have been designed or adapted to investigate and explore various internal states among individuals with intellectual disabilities. All of the approaches used, including informant based measures, observational or experimental designs, choice paradigms, event-related potential work and neuroimaging studies have demonstrated potential methods of assessment with robust reliability and validity and the understanding of gene-brain-behavior pathways for each specific syndrome.

In relation to emotion and internal states, investigating syndrome specific differences suggests support for underlying genetic mechanisms that determine these specific pathways. This highlights the importance of considering the etiology of an individual's intellectual disability when considering internal and emotional states and experiences. For example, while eye contact may be highly potent and evoke laughing and smiling in individuals with Angelman syndrome, this finding cannot be generalized to the wider population of individuals with intellectual disabilities as some individuals may find it less reinforcing or even uncomfortable (e.g., those with autism or Fragile-X (e.g., Knivsberg, Reichelt, Hoien, & Nodland, 2002)). There is no reason to suggest that this interplay between genetics and expression of internal states should be unique to the syndromes discussed within this review, and indeed, there is emerging literature of increased internal states within other genetic syndromes associated with severe or profound intellectual disabilities (e.g., Cornelia de Lange syndrome; Richards et al., 2009).

Overall, it is apparent that the knowledge and literature base is at its peak in areas where there has been integration within and across methodologies and subject areas such as anxiety, depression, pain and the syndrome-specific literature. It, therefore, is imperative that research within the area of emotional expression in individuals with severe or profound intellectual disabilities strives not only to enhance knowledge but also to inform and inspire the use of multiple approaches. We will not be successful at understanding and intervening in complex problems or behaviors until we begin to generate multilevel models (Oliver & Woodcock, 2008). Only when this has been established can behavior begin to be understood in terms of its association with the environment, the brain and ultimately, the individual's genetics. This level of understanding can then promote person-centred, empirically supported interventions that recognize the importance of emotional and internal states at an individual level.

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