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Psychological Profiles Associated with Prader-Willi Syndrome

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Prader-Willi syndrome (PWS) is a genetic disorder caused by an anomaly on chromosome 15 in the q11-13 region. Originally, it was diagnosed by clinical criteria agreed upon in 1993 (Holm et al., 1993), but now it is confirmed by genetic testing and the clinical criteria are used to prompt genetic testing for PWS (Gunay-Aygun, Schwartz, Heeger, O’Riordan, & Cassidy, 2001; McCandless & Cassidy, 2006). PWS is characterized by hypotonia at birth, obesity due to hyperphagia which begins in early childhood, abnormalities with the hypothalamus (with results such as short stature and hypogonadism), mild to moderate cognitive impairment, and a variety of maladaptive behaviors described as tantrums, stubbornness, and obsessive compulsive behavior (Butler, Hanchett, & Thompson, 2006; Holm et al., 1993; Whittington & Holland, 2004).

As the first example of gene imprinting (i.e., gene expression is dependent upon the parent of origin) in humans, PWS has been and continues to be extensively studied (Dykens et al., 2000). In a typical human body, the genes in the q11-13 region on chromosome 15 are not expressed on the maternal chromosome; hence, only the genes on the paternal chromosome 15 in the q11-13 region show active expression (Soni et al., 2007). It is when genes in this region are not expressed on the paternal chromosome, that Prader-Willi syndrome results (Soni et al., 2007). This lack of expression typically occurs in one of two ways: approximately 70% of the cases of PWS are due to a deletion of the q11-13 region on the paternal chromosome and approximately 25% of the cases occur due to maternal uniparental disomy (UPD), which occurs when the maternal chromosome 15 copies fail to separate resulting in two copies of the maternal chromosome 15 being passed on while none from the paternal chromosome 15 are (Hartley et al., 2005; Soni et al., 2007). The remaining small minority of cases of PWS result from either a translocation of the material from region q11-13 on the paternal chromosome (resulting in an

expression similar to a deletion) or from a defect in the imprinting center so that the genes at region q11-13 on the paternal chromosome are not expressed (resulting in functional expression identical to a UPD) (Buiting & Horsthemke, 2006; Soni et al., 2007).

As the methods for genetic testing have become more precise, increasing numbers of studies have assessed differences among people with PWS with different genetic subtypes (e.g., Cassidy et al., 1997; Dykens & Roof, 2008; Hartley, MacLean, Butler, Zarcone, & Thompson, 2005; Milner et al., 2005; Webb et al., 2002; Whittington & Holland, 2004). Earlier studies were only able to compare those who have PWS because of paternal deletion to those who have PWS because of maternal uniparental disomy, while more recent studies (beginning with Butler et al., 2004) were able to additionally look at differences between people who have the deletion Type 1 (TI) form and the deletion Type 2 (TII) form. A TI deletion has a breakpoint nearer the center of chromosome 15 than does a TII deletion, which results in a loss of more genetic material, approximately 500 kb (Butler et al., 2004; Hartley et al., 2005). Some research has shown differences in specific areas of behavior and cognitive abilities among those with different genetic subtypes of PWS (Dykens & Roof, 2008). The reason that this research into phenotypic differences among genotypes is so important is that it “may reveal genes causing specific clinical manifestations or enhance understanding of the impact of imprinting on genotype and phenotype studies in PWS” (Roof et al., 2000, p. 26). Clearly, the more that is understood about the causes of a particular behavior, the more effective and efficient the designs of any related interventions can be.

Generally, PWS is not considered to be inherited though Percy et al. (2007) reported that occasionally PWS is transmitted through autosomal dominance. In her review of the literature, Kundert (2008) found the birth prevalence of PWS to be 1:8,000-29,000 and the population

prevalence to be 1:52,000-76,000. As these data imply, early mortality has been a characteristic of PWS due to complications from obesity and hypotonia (e.g., cardiovascular and respiratory disorders) (Eiholzer & Lee, 2006) but with improving treatments such as weight management, exercise, and growth hormone, the average length of life may be increasing.

The purpose of this paper is to present an overview of the psychological characteristics associated with PWS. Because psychological characteristics are by definition less visible than physical characteristics, they may be more easily hidden or misinterpreted. The aim of this paper is to provide a context for the visible behaviors that one might observe in someone who has Prader-Willi syndrome. For discussions of physical and medical considerations including hyperphagia and related behaviors in PWS, the reader is referred to the research and reviews reported by Eiholzer and Lee (2006), Goelz (2006), Scheimann, Lee, and Ellis (2006), and Whitman and Jackson (2006).

Academic and Social Cognitive Functioning

PWS usually co-occurs with intellectual disabilities (ID), typically in the mild to moderate range (Holm et al., 1993). Whittington et al. (2004b) found that there is a bell curve distribution of full scale IQ shifted 40 points downward from the general population IQ, which suggests that PWS has a “global, rather than specific effect on cognition” (p. 182). However, relative strengths in reading as opposed to arithmetic have been noted when compared to others with ID (Whittington et al., 2004a) and people with PWS tend to be better at visual learning than auditory learning (Chedd, Levine, & Wharton, 2006; Walley & Donaldson, 2005).

Impairments in “social cognition, literal mindedness and cognitive inflexibility are also present” (Whittington et al., 2004b, p. 172). These characteristics of PWS are “strikingly similar

to the symptoms characteristic of frontal dysfunction in lesional pathologies and autism spectrum disorders” (Juaregi et al., 2007, p. 252). Hence, this cognitive profile has been studied in terms of executive function deficits, which are regulated in the frontal lobes (Juaregi et al., 2007; Walley & Donaldson, 2005), social competence (Koenig, Klin, & Schultz, 2004; Rosner, Hodapp, Fidler, Sagun, & Dykens, 2004), and comparison with characteristics associated with autism (Greaves, Prince, Evans, & Charman, 2006; Koenig et al., 2004). Different conclusions have been reached as to whether an actual impairment in executive function exists (Juaregi et al., 2007) or whether the impairment instead is in the phonological loop¹, which is located in the left parietal lobe (Walley & Donaldson, 2005). However, research is in agreement that people with PWS tend to have lower social competence than IQ-matched peers. For example, Rosner et al. (2004) found that a PWS group rated below average more often than both a Down syndrome group and a Williams syndrome group when compared using the social competence domain of the Child Behavior Checklist. In another study to measure social functioning, Koenig et al. (2004) found that a group of people with PWS and a group of people with pervasive developmental disorder (PDD) performed similarly and both performed more poorly than an IQ-matched group on tests to recognize the relevant social aspects of a visual display.

In addition to deficits in social ability, people with PWS often function at an academic level below that predicted by their IQ (Whittington et al., 2004a). Maladaptive or impaired social behaviors (e.g. immature social behavior, tantrums, rigid thinking, and obsessive-compulsive behavior) have been cited as reasons for adaptive functioning below what is expected from IQ scores (Dykens et al., 2000; Whitman & Jackson, 2006; Whittington et al., 2004a). Additionally, although IQs may be high, problems with “understanding and using language have [a further] negative impact on cognitive functioning. Studies document deficits in

vocabulary, receptive and expressive language, language comprehension, pragmatic language, poor discourse and conversational skills, and shortened length of utterances” (Whitman & Jackson, 2006, p. 322).

Regarding academic and social cognitive functioning, two main differences have been documented between persons with a UPD cause of PWS and persons with a deletion cause of PWS. First, people with a UPD have been found to score significantly higher in verbal IQ than do those with a deletion (Roof et al., 2000; Whittington et al., 2004b; Zarcone et al., 2007). More specifically, Roof et al. (2000) found that individuals with UPD scored significantly higher in their verbal IQ than their performance IQ while individuals with deletion scored higher on their performance IQ than their verbal IQ; the performance scores themselves were not significantly different. Roof et al. (2000) also found that the mean average verbal IQ for the UPD group was 69.9 while it was 60.8 for the deletion group. Since these scores would put the UPD group on the borderline for a classification of ID while the deletion group would be solidly within the classification based on the intellectual functioning criterion, Roof et al. (2000) pointed out that this finding “carries substantial implications for individual classification, placement, and service decisions” (p. 28).

A second difference is that those with a UPD are more likely to have co-occurrence of autism (a disorder in which impairment in social functioning is a prime characteristic) than those with a deletion (Dykens et al., 2000). Milner et al. (2005) found their UPD group to have “increased autistic-like social impairments” when compared to those with a deletion even though the two groups performed similarly in terms of adaptive social behavior and were matched for full scale IQ (p. 1094). However, Dykens et al. (2000) stressed that as a whole, “Prader-Willi

syndrome does not appear to include a heightened risk of autism spectrum disorder, beyond the risk due to [ID]” (p. 194).

Effective teaching methods take into account the tendency toward characteristics associated with frontal lobe dysfunction. “The frontal lobes control personality and expression of emotion, motivation, and initiation, as well as inhibit an inappropriate or impulsive action. They store information and affect a person’s ability to concentrate, plan strategically, and think in abstract terms” (MacKay & Percy, 2007, p. 114). Whittington et al. (2004a) have recommended “teaching based on concrete materials, rule-based learning and visual methods [since] people with PWS [typically have] poor auditory short-term memory, difficulty in generalizing and do not understand abstract concepts” (p. 199). Basic social skills training such as “turn taking and perspective training” is recommended (Dykens & Shah, 2003). Whitman and Jackson (2006) gave a thorough list of practical strategies to use specifically for the characteristics of “rigid thought process and inability to be behaviorally flexible,” “perseverative or obsessive thinking,” and “tenuous emotional control or short frustration tolerance” (pp. 332-334).

Obsessive Compulsive Behavior

“Obsessive/compulsive behavior,” “rigid,” and “perseverating” characteristics are included in the minor criteria of Holm et al.’s (1993, p. 399) diagnostic criteria. However, researchers are not yet in agreement as to whether these and other characteristics commonly observed in people who have PWS constitute clinical Obsessive Compulsive Disorder (OCD) or whether the characteristics are unique (Whitman & Thompson, 2006; Wigren & Hansen, 2003). One problem is that the obsessive component of OCD is not clearly documented, though this may be due to the fact that persons who have PWS often have difficulty in “assessing and

expressing the presence of the internalizing components of obsessions” (Whitman & Thompson, 2006, p. 262). Even though the compulsions exhibited by people with PWS may be similar to those with OCD (Zarcone et al., 2007), the clinical criteria for OCD might not be met.

Whitman and Thompson (2006) found that some researchers argue that these obsessive compulsive or ritualistic behaviors appear at the same developmental stage as they do in typically developing children. Moreover, they stated, one of the criteria for classic OCD is that the behaviors are felt by the person exhibiting them to be abnormal or psychologically stressful, yet the children with PWS do not view their behaviors as out of the range of normal or as stressful. Rather, Whitman and Thompson (2006) proposed, it is the caregivers, not the children, who are stressed by the behaviors. Dykens et al. (1999), however, found that individuals with PWS did in fact have significant stress around their compulsions.

Other studies also concluded that these obsessive compulsive symptoms were not merely developmental phases. For instance, Dimitropoulos, Feurer, Butler, & Thompson (2001) found statistically significant differences in the compulsive behaviors of children ages 2-5 among a group who had PWS, who had Down Syndrome and who were typically developing, and Webb et al. (2002) found statistically significant differences in obsessive compulsive behaviors between a group of people with genetically diagnosed PWS and a PWS-like group (i.e., people who were thought to have PWS but, upon genetic testing, determined to not have it). As Wigren and Hansen (2003) concluded, “differentiation between delayed childhood rituals and pathological manifestations of compulsive features is complex in PWS populations” (p. 428).

Yet another viewpoint is that these obsessive compulsive characteristics have more in common with autism spectrum disorders (ASD) than with OCD (Whitman & Thompson, 2006). This idea is in line with similarities noted previously between the cognitive profiles of people

with PWS and ASD. For example, the temper tantrums and repetitive questions characteristic in the PWS profile may be linked to resistance to change or rigidity (Woodcock, Oliver, & Humphreys, 2008).

Because there are so many ways to approach a study of rigid, perseverative, obsessive compulsive and related behaviors, many different assessment methods and definitions have been used (e.g., Butler, Bittel, Kibiryeve, Talebizadeh, & Thompson, 2004; Didden, Korzilius, & Curfs, 2007; Dykens, 2004; Dykens, Cassidy, & King, 1999; Greaves et al., 2006; Reddy & Pfeiffer, 2007; Zarcone et al., 2007). Because of this variance, some inconsistencies in the conclusions reached by different research may be due to different definitions of obsessive compulsive behavior, different standardized assessment tests, different age groups of participants and different divisions into genetic subgroups (i.e., no distinction, distinction between deletion and UPD, or distinction among TI deletion, TII deletion, and UPD groups). Some of the behaviors classified in studies as obsessive compulsive are described as hoarding, checking, arranging, repetitive, just right, cleanliness, obsessive rumination, stereotyped behavior, and skin-picking (Dimitropoulos et al., 2001; Dykens, 2004; Juaregi et al., 2007; Wigren & Hansen, 2003).

Among the variously described obsessive or compulsive behaviors, skin-picking is unique. Holms et al. (1993) lists skin picking as a stand-alone minor criterion for PWS beside the minor criterion of “characteristic behavior problems” (which includes obsessive/compulsive behavior and others; p. 399). Hence, skin-picking is incorporated into studies in varied ways: as a symptom or representation of obsessive compulsive behavior; as a symptom or representation of self-injurious behavior; as one of many maladaptive behaviors; or is studied independently (Butler, et al., 2004; Didden et al., 2007; Dykens, Cassidy, & King, 1999; Dykens, Hodapp, &

Finucane, 2000). Vythilingum and Stein (2008) reported on the incidence of OCD in dermatology clinics and described the overlapping relationship between OCD, self-injurious behavior, and skin-picking in the general population:

Unlike OCD, compulsive picking may have an early or late onset, and is more prevalent in females. Comorbidity with other psychiatric disorders is high, notably with OCD, [obsessive compulsive spectrum disorders], mood disorders, and substance use disorders...It is possible to differentiate [compulsive skin-picking] from other self-injurious behavior...[S]elf-injury is usually impulsive and is associated with suicidal ideation, dissociation, and aggression; this is in contrast to skin picking, which is often a compulsive act performed to relieve tension. (p. 105)

Skin-picking by people with PWS is quite common and has been found to have a moderately positive correlation with severity of compulsive behaviors but not with other self-injurious behavior, level of ID, residence, psychosis, or gender (Didden et al., 2007). Three studies have found skin-picking and other maladaptive compulsive behaviors to be more severe among people in their 20s as compared to children, adolescents and older adults (Clarke, Boer, Chung, Sturmey, & Webb 1996; Dykens, 2004; Hartley, MacLean, Butler, Zarcone, & Thompson, 2005). Dykens (2004) also found that girls were more likely to skin-pick than boys, which matches the gender bias observed in skin-picking in the general population (Vythilingum & Stein, 2008).

Although some studies have found an inverse relationship between Body Mass Index (BMI) and skin-picking or other compulsive behaviors such as hoarding, a common hypothesis for this association is that the stress involved in low-weight maintenance routines may increase

anxiety and hence, anxiety-reducing coping mechanisms such as compulsions (Dykens, 2004). Dykens (2004) discussed another hypothesis: the hormone ghrelin is an appetite stimulant and is found in elevated levels in persons with PWS and the levels remain high even after food consumption. Assuming that eating provides some measure of psychological relief, in that the appetite is continuously stimulated and eating would be answering that drive, and that thinner people consume less food, thinner people will experience less relief (Dykens, 2004).

As mentioned previously, obsessive compulsive behaviors have been studied with regard to similarities with behaviors associated with ASD (Juaregi et al., 2007). Greaves et al. (2006) compared levels of repetitive and ritualistic behaviors between two groups of children: one who had PWS and one who had autism. They found that out of 19 categories on the Childhood Routines Inventory, the two groups scored similarly on 15 of them. Of the remaining four categories, the PWS group rated higher in the “collects and stores items” category but rated significantly lower in the categories of “line up objects,” “aware of detail at home,” and “strong preference for certain foods” (p. 97). The low rating in this latter category would certainly be expected given that hyperphagia is universal among people with PWS. The authors also found that in both groups repetitive behavior decreased as developmental level increased, which seems to add evidence to the research which Whitman and Thompson (2006) described regarding compulsions as a typical developmental stage. However, the decrease in maladaptive behaviors in childhood due to developmental phases does not explain the significant spike in maladaptive behaviors that was observed in early adulthood by Clarke, Boer, Chung, Sturmey, and Webb (1996), Dykens (2004), and Hartley, MacLean, Butler, Zarcone, and Thompson (2005).

The research on differences in obsessive compulsive symptoms among PWS subtypes is inconclusive. For example, some studies have found a significantly greater number of people who skin-picked in a deletion group versus a UPD group (Butler, Bittel, Kibiryeva, Talebizadeh, & Thompson, 2004; Dykens et al. 1999) while others have found no such distinctions (Didden et al., 2007; Webb et al., 2002). Two studies comparing deletion and UPD subgroups found similar profiles of compulsive behavior (Didden et al., 2007) and repetitive behavior (Greaves et al., 2006), but when Dykens et al. (1999) assessed obsessive compulsive behaviors overall, they were found to be a greater problem in the deletion subgroup. When the deletion subgroups TI and TII have been also been assessed for rates of compulsive behavior, some studies have found higher rates in the TI group than either TII or UPD groups (Butler et al., 2004; Zarcone et al., 2007) and other research has found similar rates (Milner et al., 2005).

The research on differences among genotypes is more consistent with regard to the obsessive compulsive behaviors that mimic those commonly observed with ASD. Similarities with characteristics of autism have been observed with higher frequency in people with the UPD form of Prader-Willi syndrome. People with UPD (compared to those with a TI or TII) were found to be “more likely to return to their compulsions when interrupted which is similar to the repetitive behaviour associated with autism” (Zarcone et al., 2007, p.485). Butler et al. (2004) found similar results except that they found the TI group was aligned with the UPD group. People with UPD have been found to have a higher risk for autistic-like symptomology (Milner et al., 2005) and Dykens et al. (1999) observed that individuals who had a co-occurrence of PWS and autism tended to have the UPD form of PWS.

Both pharmacological and behavioral interventions have been used for obsessive compulsive behaviors exhibited by individuals with PWS. Dykens et al. (1999) and

Dimitropoulos et al. (2001) summarized research which suggests that oxytocin and serotonin levels may affect certain OCD behaviors since these chemicals mediate certain grooming behaviors, a common category of compulsive behaviors. For example, Zarcone et al. (2007) found that individuals in a TI group tended to have compulsions related to personal cleanliness (e.g., grooming) while those in the TII group tended to have compulsions related to academic areas (e.g. counting); therefore, medications affecting serotonin levels may be more effective with people with TI (grooming compulsions) than TII (non-grooming compulsions). Research focused on adjusting serotonin levels has reported successes with using serotonergic reuptake inhibitors to reduce stereotyped behaviors in children with autism and repetitive behaviors in people with PWS (Greaves et al., 2006) but more research on larger populations with consistent results is needed (Dykens & Shah, 2003). In a study to determine the usefulness of the antiepileptic drug Topiramate in reducing appetite in people with PWS, Shapira, Lessig, Lewis, Goodman, & Driscoll (2004) found the unexpected result that skin-picking was reduced; although the study size was small (n=8), further study might reveal this as an effective treatment for skin-picking.

In light of the absence of a reliable treatment for skin-picking, which is an overwhelmingly common compulsive behavior among people with PWS, Didden et al. (2007) emphasized the need to determine the function of skin-picking in order to increase the likelihood that the treatment will be effective. In their study, the function of skin-picking was most often found to be tension or arousal reduction so, in cases like that, treatment might include “relaxation training, response prevention, habit reversal training, anxiety and anger management and/or teaching coping strategies in how to deal with psychological stressors” (p. 417).

Just as Didden et al. (2007) suggested for skin-picking, determining the function of any repetitive or compulsive behavior will guide the choice of an intervention which will address the need being met by the behavior. Predicting a maladaptive behavior by identifying the triggers and the function of the behavior, and, based on this information, making changes to the environment or adding supports or skills so that the behavior of concern is no longer necessary, are tenets of positive behavior support and based on applied behavior analysis. Dykens and Shah (2003) give an example of applying these principles:

[A]s a result of their compulsivity, many people with Prader-Willi syndrome have difficulty with unexpected changes or with the transition from one activity or thought to the next. They may thus need extra help getting 'unstuck' from a particular thought or behavior. It may be helpful for parents or teachers to use special visual or auditory transition cues, to give ample warning before a transition or to develop a behavioural programme for appropriate transitions. (p.173)

Behavioral interventions to treat obsessive compulsive behaviors in people with autism have been successful (Greaves et al., 2006) and could presumably translate to the PWS community and Zarcone et al. (2007) suggested cognitive behavioral therapy for persons with a TI deletion in particular.

Psychopathology

Although psychiatric illnesses (i.e., anxiety, depression, mood disorders, and psychotic disorders) are not found on the list of consensus diagnostic criteria (Holm et al., 1993), surveys of the literature reveal a higher incidence of psychopathology in individuals with PWS when compared to both the general population and to other people with ID (Dykens et al., 2000;

Kundert, 2008; Reddy & Pfeiffer, 2007). Specifically, the risk of clinical psychosis or an affective disorder increases with age, especially in adulthood (Boer et al., 2002; Didden, 2007; Soni, 2007). But, interestingly, when clinical psychosis and affective disorders are excluded, people with PWS may actually experience a decrease in milder psychiatric problems such as anxiety and depression as they age past their twenties, which is in contrast to other syndromes such as Down syndrome and ID in general (Dykens, 2004).

Like obsessive compulsive behavior and related behaviors, psychopathology has been defined in studies in varied ways, sometimes with strict clinical criteria to meet clinical psychosis or depressive conditions and at other times it has been surmised from maladaptive behavior. Accordingly, the assessments used have varied widely in their focus and clinical criteria. For example, Dykens (2004) used the Childhood Behavior Checklist to assess, among other scales, general anxiety and depression across different age groups. Meanwhile, Soni et al. (2007) used much stricter clinical criteria and excluded from their study “[i]ndividuals with only a history of behavior problems, obsessive-compulsive symptoms, [and] mood swings not meeting [International Statistical Classification of Diseases and Related Health Problems Tenth Revision] diagnostic criteria for a mood disorder, or temper tantrums typical of PWS” (p. 33).

Of particular note regarding psychopathology and PWS is that individuals with UPD have an extremely high risk of developing a psychotic disorder later in life, i.e., late 20s to early 30s (Boer et al., 2002; Soni et al., 2007). Since this risk is so high, plans may be designed ahead of time to deal with the situation when it arises. With increased preparation time, better supports should be able to be put in place, for the individual as well as her/his family. Persons with UPD vs. a deletion also seem to be affected more severely by their psychiatric illnesses as indicated by

a higher frequency and number of psychiatric episodes and by having tried, on average, a greater number of medications (Soni et al., 2007)

Soni et al. (2007) evaluated treatments that their study group of 46 adults with psychiatric illness had received over their lifetimes and found that no one reported ever having received any formal psychotherapy, including cognitive-behavioral therapy. Psychotropic medications were not only overwhelmingly prescribed for those with psychopathologies but had also been prescribed at some point for 36% of the 73 individuals who had *not* met the study-defined criteria for psychiatric illness. The psychotropic medications had mostly been prescribed for conditions labeled challenging behavior, mood swings and temper tantrums. An important finding of the study that the authors noted warrants further research is that individuals who were taking a mood-stabilizing medication were more likely to have a recurrence of the psychiatric episode. Soni et al. (2007) hypothesized that the results of their evaluations suggest that mood-stabilizing medications which act on GABA receptors may be problematic whereas ones that do not, such as lithium, may be effective. One of the confounding factors in studying the uses of psychotic medications is that “due to an altered body composition and metabolism, persons with Prader-Willi syndrome often have extremely idiosyncratic, frequently unpredictable, and often negative responses to medications” (Whitman & Jackson, 2006, p. 339).

Discussion

To review, persons with Prader-Willi syndrome have a psychological profile of mild to moderate ID, academic and social functioning below that predicted by IQ scores, rigidity in thought processes, strong preference for routines, better ability to learn visually than aurally (especially for those with UPD), and a tendency toward compulsions, particularly skin-picking

(Butler et al., 2006; Rosner et al., 2004; Whittington et al., 2004a, 2004b). Breaking down the psychological profile into genetic subtypes, a few tendencies appear. Persons with deletion tend to have lower IQ scores (particularly verbal scores) than persons with UPD but greater social competence when matched by IQ scores (Milner et al., 2005; Roof et al., 2000). Those with a co-occurrence of autism or who demonstrate social impairments, like those associated with autism, usually have the UPD form of PWS, though the rate of occurrence of autism is not significantly higher in the PWS population than in the general ID population (Dykens et al., 1999). Individuals with UPD have very high rates of some form of psychosis in adulthood after their 20s, while milder forms of psychiatric illnesses such as depression or obsessive compulsive disorder seem to abate in adults in their 30's and 40's, counter to the trend in the general population of people with ID (Dykens, 2004; Soni et al., 2007).

This paper has reviewed general information about the psychological profile of people with Prader-Willi but it has cited almost exclusively literature about people who have not received growth hormone (GH) therapy. In 1998 growth hormone therapy was approved by the FDA as a treatment for adults with a growth hormone deficiency and in 2000 it was approved as treatment for children with PWS (Carrel, Lee, & Mogul, 2006). As a result, research is beginning to be published on GH treatments and results show that people receiving GH therapy may exhibit significantly different profiles than those who have not received GH treatments. Two studies have found that adults receiving GH therapy significantly improved not only in physical composition and performance but also in mental speed, mental flexibility, anxiety, depression, positive well-being (when rated by persons with PWS), self control (when rated by parents), and general health (Bertella et al., 2007; Höybe, Thorén, & Böhm, 2005).

Optimal quality of life is the goal of any intervention, whether for Prader-Willi syndrome or a fractured bone. In order to know whether that goal has been reached, a measure of success is needed. Dykens, Schwenk, Maxwell, & Myatt (2007) have proposed that the Sentence Completion and Three Wish tasks might be effective tools to measure self-perception and perception of the world in individuals with ID, including those with PWS. These tasks are “semi-projective” assessments so that the individual with ID is given some structure yet the response is “open-ended” (p. 589). Individuals are asked to finish a sentence that the examiner begins, in one of the tasks, and in the other task, the individual is asked for 3 magic wishes. Measures of perception would translate into measures of quality of life. Besides our professional observations and educated guesses on the psychological states (i.e. quality of life) of our clients with Prader-Willi syndrome, we could add insights from the individuals themselves, which is optimal. For instance, Dykens et al. (2007) found that persons with PWS had an unexpectedly large number of non-food thoughts. Findings like these will guide interventions and goals more effectively and meaningfully. The outlook for people with Prader-Willi is more positive than ever considering the continually expanding body of knowledge of specific gene functions, the growing emphasis on/demand for interventions based on function, the opportunity to piggy-back off of the currently massive interest in and research into behaviors associated with autism that are also associated with PWS, and the increasing availability of growth hormone treatments.

Footnotes

¹The phonological loop processes aural information received in working memory (phonological loop, 2009).

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