BRIEF REPORT: THE RELATIONSHIP BETWEEN COMPONENTS OF THE BEHAVIOURAL PHENOTYPE IN PRADER-WILLI SYNDROME[1]

Running title:
Temper outburst precursors in Prader-Willi syndrome

Key Words:

Temper tantrum, repetitive questions, stubbornness, precursor behaviours, functional equivalence, behavioural hierarchy.

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ABSTRACT

Background

Repetitive questions and temper outbursts form part of the behavioural phenotype of Prader-Willi syndrome (PWS). We investigated the phenomenology of temper outbursts in PWS and their relationship with other PWS behavioural characteristics.

Method

Four individuals with PWS were observed (5-10 hours), during a number of experimental and natural environment challenges, some of which were expected to trigger temper outbursts. Individual behaviours including crying, ignoring, arguing, questioning, stereotypy, frowning and posture changes were recorded and subjected to lag sequential analysis.

Results

All participants were significantly more likely to show repetitive questioning before more challenging behaviours such as crying, arguing or ignoring requests. Precursor behaviours such as frowning and stereotypical behaviour were identified in three participants.

Conclusions

Temper outbursts in PWS may be associated with other PWS behavioural phenotypic characteristics such as repetitive questions and 'stubbornness'. A progression of behaviours may lead up to the most challenging temper outburst behaviours. This may have important implications for effective coping strategies.

INTRODUCTION

Prader-Willi syndrome (PWS, prevalence: 1:10,000 to 1: 25,000) is caused by absence of paternal genetic information in the chromosome 15 q11-q13 region and is usually associated with mild to moderate intellectual disability (e.g. Whittington et al., 2004). The PWS behavioural phenotype includes excessive eating, specific repetitive and self-injurious behaviours, temper outbursts, mood disturbance, repetitive questioning, 'stubbornness', 'disobedience', excessive daytime sleepiness and under activity (e.g. Einfield, Smith, Durvasula, Florio & Tonge, 1999; Richdale, Cotton & Hibbit, 1999; Holland et al., 2003; Arron, Oliver, Berg, Moss, & Burbidge, in review; Oliver, Berg, Burbidge, Arron & Moss, in review; Moss, Oliver, Arron, Burbidge, & Berg, in review).

A number of different structured interviews and questionnaires have been used to document more frequent and severe temper outbursts (also referred to as 'temper tantrums') in individuals with PWS when compared to groups of individuals with intellectual disabilities of mixed and unknown aetiologies, individuals with other genetic syndromes, or typically developing children (Clarke, Boer, Chung, Sturmey & Webb, 1996; Dimitropoulos, Feurer, Butler & Thompson, 2001; Einfield et al., 1999; Waltz & Benson, 2002; Holland et al., 2003). Holland et al. used factor analysis to show that temper outbursts in PWS appeared to be associated with repetitive questions and obsessive, possessive and violent/aggressive behaviour, but no direct observations were made to support this analysis. To date, no studies have directly examined the phenomenology of temper outbursts in PWS or the relationship between components of the behavioural phenotype within individuals with PWS.

Description of the precursors to and phenomenology of temper outbursts in PWS might be beneficial for intervention. Precursor behaviours can sometimes reliably precede a specific challenging behaviour and share the same function (e.g. Smith & Churchill, 2002). Identifying precursor behaviours of temper outbursts in PWS could assist in the development of effective coping strategies by allowing functionally equivalent behaviours to be introduced into the repertoire and reinforced when precursors are shown, hence reducing the likelihood of a temper outburst.

As part of a larger study into the relationship between specific cognitive impairments and temper outbursts in PWS, we collected observational data on the temper outbursts shown by four individuals with PWS (Woodcock, Oliver and Humphreys, in review). The aim of the current report is to describe the specific behaviours that comprise the outbursts, behaviours that are precursors to the outbursts and associations between temper outbursts, repetitive questions and 'disobedience' and 'stubbornness'.

METHOD

Participants

Four females (RB, AH, AG and KT) with a paternal deletion in the chromosome 15 q11-q13 region participated[2]. Participants' ages at the beginning of the study were 14:2, 14:3, 12:6 and 17:1 years. Approximately one year before the beginning of the study Similarities, Vocabulary, Block design and Object assembly subtests from the Wechsler Intelligence Scales for Children (WISC III; Wechsler, Golombok & Rust, 1992) were administered. At this time the participants showed test age equivalents of between less than 6:2 years and 9:6 years.

[2] Genetic characterisation was based on parental report of previous genetic tests that had been carried out at a hospital or by a genetic counselling service.

Measures

Parents and carers were asked to report freely the behaviours that lead up to and comprise a temper outburst shown by the child they care for. As part of a larger study investigating the causes of temper outbursts in PWS, participants were observed whilst engaged in series of cognitive challenges designed to vary the level of demand placed on attention switching as this is thought to be associated with temper outbursts (see Woodcock, Oliver & Humphreys, in review). Participants were also observed whilst playing experimentally designed tabletop games that manipulated specific environmental conditions (degree of predictability) potentially associated with temper outbursts. Finally, participants were observed in their natural environment, during which time manipulations (deviations from routine or expectations) were imposed that, based on reports, were expected to trigger temper outbursts.

Procedure

Participants were observed over a period of between five and nine weeks; between 100 and 145 minutes whilst engaged in cognitive challenges, between 100 and 138 minutes whilst playing tabletop games and between 80 and 358 minutes in their natural environments.

Behaviour Observation

All observation periods were recorded using a video camera. Two observers independently coded 10% of the footage and Kappa inter-observer reliability indices were calculated based on successive 5s intervals for behaviours shown by each participant. Kappa indices were: questioning (questions pertinent to the current situation): .66 to .82; arguing (statement of disagreement, giving orders, making

demands, or refusing requests): .56 to .76; crying (crying with tears or speech or non-speech vocalisations associated with crying): .47 to .76; frowning by RB (lowered position of the inside ends of the eyebrows and a furrowing of the brow): .72; stereotypical behaviour by AG (inter-weaving of the fingers with hands at midline, tapping of the fingers on the forearm or non-speech vocalisation made deep within the throat): .79; stereotypical behaviour by KT (movement of the lips whilst not engaged in vocalisation): .44; ignoring requests by KT (not carrying out a direct request made by the researcher or a teacher before the request had had to be repeated): .99; posture changes by KT (movement of the whole body to result in a different sitting position, or the shoulders being raised and lowered coupled with lowering of the trunk): .66.

Data Analysis

To examine the sequence of behaviours leading up to and comprising a temper outburst, two time-based sequential lag analyses were carried out for each participant, lagging from the presence of the two most disruptive reported behaviours that were observed (crying and arguing in RB, AH and AG; ignoring and arguing in KT: criterion behaviours). Lags were examined in one-minute intervals preceding and following the presence of the criterion behaviours, and the conditional probability of the presence of other behaviours (questioning in all participants, stereotypy in AG and KT, frowning in RB and posture changes in KT) in these intervals was calculated. The analysis was restricted up to the onset of the next criterion behaviour, ensuring consideration of probabilities of behaviours surrounding a single incidence of the criterion behaviour. It was necessary to determine whether the conditional probabilities of the behaviours surrounding a criterion were greater than would be expected by chance. Yules Q values were therefore calculated and a value of 0.4

(equivalent to an Odds Ratio of 2.33; i.e. 2.33 times more likely than would be expected by chance) was taken as the point above which the behaviours were considered to be associated (e.g. Hearn, 2003). These analyses were carried out using the computer software package ObsWin32, version 3.0 (Martin, Oliver & Hall, 2000).

RESULTS

In all participants, carers reported that behaviours leading up to and comprising temper outbursts included arguing, shouting/ screaming, facial flushing, stamping, angry facial expression and specific movements of parts of the body or the whole body (including rigid posture). RB, AH and AG were reported to show crying and excessive salivating. Ignoring and throwing/ banging objects was reported in RB and KT. Additionally, AH was reported to show 'glaring' and KT was reported to ask questions and appear puzzled. The operationally defined behaviours therefore corresponded to carers' reports.

To demonstrate the results of the lag analyses, diagrams were constructed for the two criterion behaviours for each participant. Behaviours associated with the criterion (Yules Q >0.4) are shown in circles at successive time lags along a horizontal axis, with the strength of the association shown on the arrow connecting the behaviour to the central circle (the criterion behaviour). These diagrams are shown in Figure 1.

Figure 1 about here

It can be seen that in all participants, arguing was likely to be shown within the same minute as crying or ignoring (KT only), but that arguing was more likely to precede

the crying or ignoring than crying or ignoring was likely to precede arguing. In all participants, questioning was most likely to occur between two and three minutes preceding or following arguing and crying/ ignoring. The most likely sequence of behaviours for AH was therefore question, argue, cry, question. RB was most likely to show frowning before questioning, suggesting that the most likely sequence of behaviours for RB was frown, question, argue, cry. AG's stereotypical behaviour was likely to occur preceding or following arguing, as did her questioning, meaning that the most likely sequence of her behaviours was stereotypy or question, argue, cry, stereotypy or question. In KT, stereotypical behaviour was most likely to precede or follow ignoring or follow arguing, and posture changes were very likely to occur preceding or following arguing. KT's behaviours therefore fell into the sequence: posture change, question, argue, stereotypical behaviour, ignore, question, posture change.

DISCUSSION

We have described specific observable behaviours that have been shown to occur during temper outbursts or to precede those outbursts in four children with PWS. In all participants, questioning was likely to precede more challenging behaviours such as crying, arguing or ignoring requests. Individual potential precursors such as frowning or stereotypical behaviour were more likely to precede more challenging behaviours in three participants. Therefore, in these four individuals, temper outbursts involve a progression of behaviours that lead up to the most challenging behaviour. This concept of a hierarchical repertoire of functionally equivalent behaviours has been described before (e.g. Oliver, Oxener, Hearn & Hall, 2001) and

has important implications for effective coping strategies that could aim to increase the effectiveness of the non-challenging behaviours in fulfilling a particular function.

It is interesting to note that repetitive questions and 'stubbornness' (as evidenced by arguing and ignoring requests), which have been reported as components of the PWS behavioural phenotype (e.g. Holland et al., 2003; Moss et al, in review, Arron et al, in review), appear to be associated with temper outbursts in PWS. This suggests that apparently distinct aspects of a behavioural phenotype may be related and evoked by the same combination of biological, cognitive or environmental phenomena. Considering the similarities in the profile of temper outburst behaviours identified across all of our participants, future research could use a group comparison to address the question of whether a characteristic series of specific behaviours appears to comprise the PWS behavioural phenotypic temper outburst.

The case study approach that we have taken does not allow for generalisation to all individuals with PWS, or for the assessment of potential differences between temper outbursts in individuals with PWS compared to other populations. All of our participants were children and female and our findings are therefore limited by a potential age and gender bias. However, this study represents a useful first step towards describing the structural relationships between components of an established behavioural phenotype.

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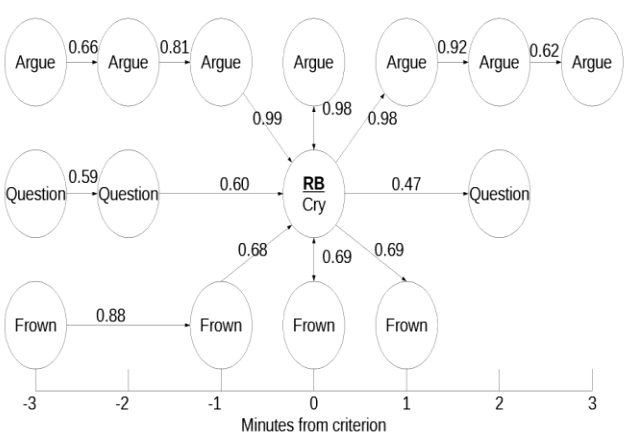
Legends

Figure 1: shows the two lag analyses conducted for each participant. Behaviours associated with each criterion (central circle in each diagram) are shown in circles connected to the criterion circle with an arrow showing the Yules Q value indicating the strength of the association between the behaviour and criterion at the specified time lag (indicated by the position of behaviours along the horizontal axis).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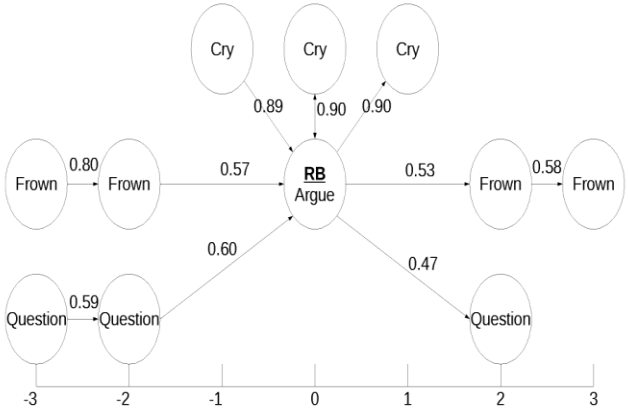
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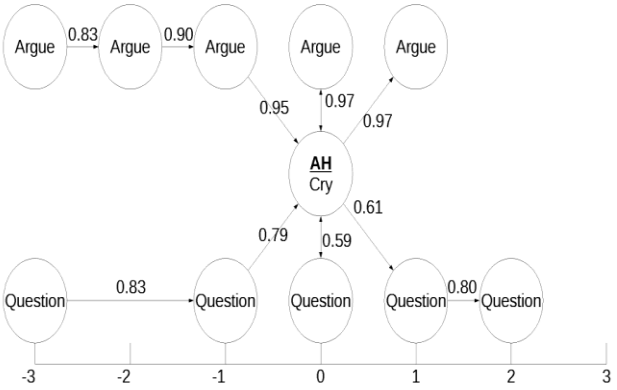
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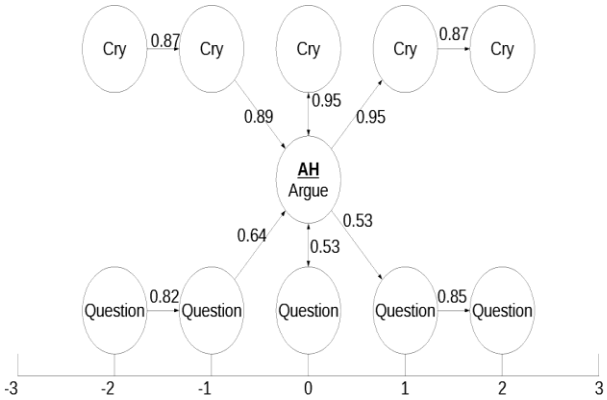
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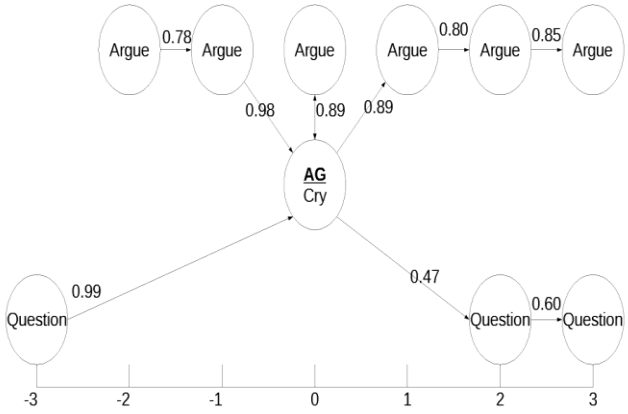
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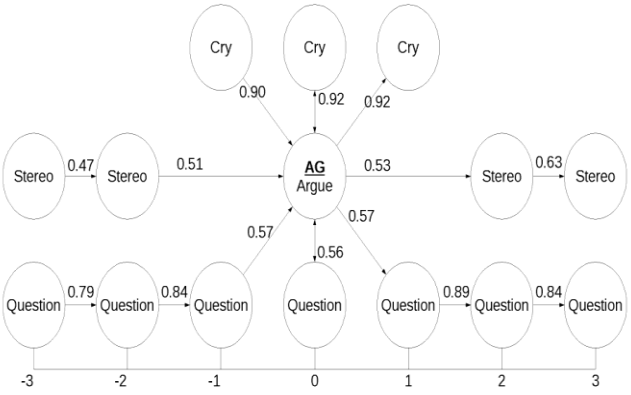
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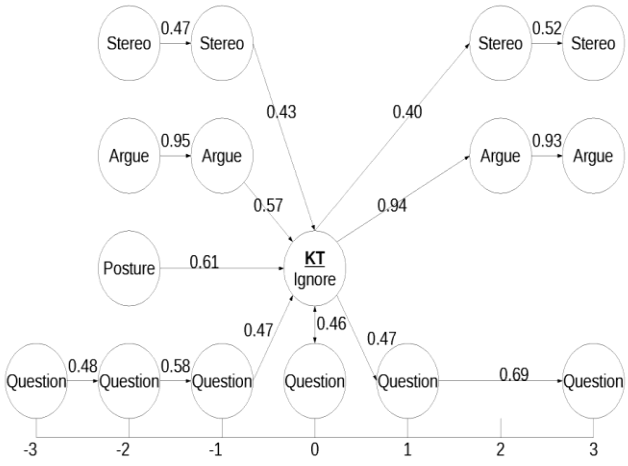
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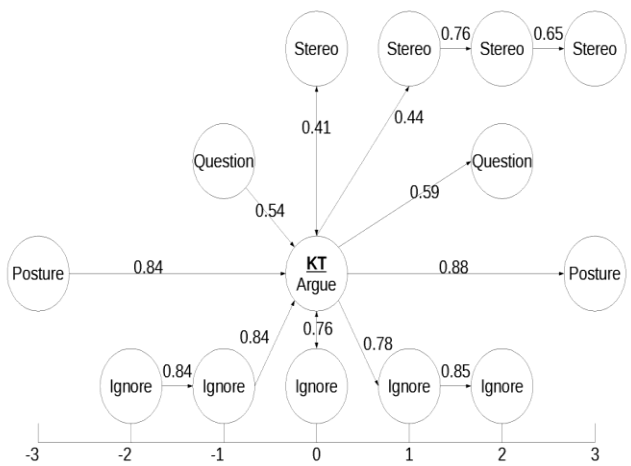
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