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Genomic imprinting and the expression of affect in Angelman syndrome: what's in the smile?

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Background: Kinship theory (or the genomic conflict hypothesis) proposes that the phenotypic effects of genomic imprinting arise from conflict between paternally and maternally inherited alleles. A prediction arising for social behaviour from this theory is that imbalance in this conflict resulting from a deletion of a maternally imprinted gene, as in Angelman syndrome (AS), will result in a behavioural phenotype that should evidence behaviours that increase access to maternally provided social resources (adult contact). **Method:** Observation of the social behaviour of children with AS ($n = 13$), caused by a deletion at 15q11-q13, and a matched comparison group ($n = 10$) was undertaken for four hours in a socially competitive setting and the effect of adult attention on child behaviours and the effect of child smiling on adult behaviours evaluated using group comparisons and observational lag sequential analyses. **Results:** The AS group smiled more than the comparison group in all settings, which had different levels of adult attention, and more when the level of adult attention was high. Smiling by children with AS evoked higher levels of adult attention, eye contact and smiling both than by chance and in comparison to other children and this effect was sustained for 30 s to 50 s. Smiling by children with AS was frequently preceded by child initiated contact toward the adult. **Discussion:** The results are consistent with a kinship theory explanation of the function of heightened levels of sociability and smiling in Angelman syndrome and provide support for an emotion signalling interpretation of the mechanism by which smiling accesses social resources. Further research on other behaviours characteristic of Angelman and Prader-Willi syndromes warrant examination from this perspective. **Keywords:** Genomic imprinting, Angelman syndrome, Prader-Willi syndrome, kinship theory, emotion signalling, behavioural phenotype, behavioural genetics, intellectual disability, prosocial behaviour. **Abbreviation:** AS: Angelman syndrome.

Genomic imprinting refers to a parent of origin effect for genes at some loci that renders a gene or genes inactive on either maternally or paternally inherited chromosomes. Angelman and Prader-Willi syndromes were the first syndromes to be identified as resulting from genetic disorders related to genomic imprinting following the observation that both disorders arose from a deletion at chromosome 15q11-q13 (Knoll et al., 1989; Cassidy, Dykens, & Williams, 2000). Angelman syndrome was observed to be caused by a deletion on the maternal chromosome; Prader-Willi syndrome by a deletion on the paternal chromosome. Although there are other causal mechanisms (uniparental disomy, imprinting defects and micro-deletions; Clayton-Smith & Laan, 2003), the effect on the phenotype is similar to a deletion, although differences are emerging (Clayton-Smith, 2001; Milner et al., 2005).

The phenotypes of Angelman and Prader-Willi syndromes differ physically, cognitively and behaviourally. Prader-Willi syndrome is characterised by short stature, small hands and feet, typical facial features (almond-shaped eyes) and hypogonadism. Hypotonia is present early with failure to thrive, followed by hyperphagia, leading to obesity if unchecked, caused by hypothalamic dysfunction resulting in impaired satiety (Holm et al., 1993). Moderate to mild intellectual disability is evident, with some cognitive strengths. High levels of compulsive behaviour and skin picking are reported (Cassidy, 1997). Angelman syndrome is characterised by dysmorphic facial appearance (pointed chin and thin upper lip, blue eyes and blond hair are common), ataxic gait, seizure disorders and distinctive EEG (Clayton-Smith, 2001). Cortical atrophy, microencephaly and ventricular dilation are evident and related to compromised expression of UBE3A in the neocortex, hippocampus and striatum and Purkinje cells in the cerebellum. UBE3A is thought to play a role in determining axon guidance, neuronal connectivity and degradation of potentially harmful proteins (Schumacher, 2001). Resultant profound or severe intellectual disability is the norm, speech rarely develops and sleep disturbance, hyperactivity, excessive sociability and preference for water play are reported (Miano et al., 2004; Clarke & Marston, 2000; Ishmael, Begleiter, & Butler, 2002). Most notably excessive laughing and smiling are common (Clayton-Smith, 2001; Horsler & Oliver, 2006a).

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Haig and Wharton (2003) have developed kinship theory (or the genomic conflict hypothesis) to account for genomic imprinting and propose that maternal and paternal alleles can be expressed differently to favour perpetuation of maternal or paternal chromosomes. This leads to a prediction that imprinted genes might be involved in influencing resource allocation for offspring (Haig & Westoby, 1989), with imprinted paternal alleles increasing the probability that maternal resources will be allocated to an offspring whilst maternal alleles have the opposite effect. Thus, the effect of imprinting is to favour perpetuation of the paternal or maternal chromosomal line by, respectively, increasing the probability of survival of the foetus and neonate, thus ensuring survival of the paternal line, or of the mother, thus facilitating further births and a higher chance of survival for the maternal line. Support for this prediction is derived from studies of imprinted genes in mice on placental function (and thus foetal growth) and neonate suckling (Isles, Davis, & Wilkinson, 2006). With regard to human behaviour perhaps the most important demonstration has been the seminal work of Skuse and colleagues on Turner's syndrome (Skuse, 2000). Goos and Silverman's (2001) interpretation of this work is that paternally active genes are primarily involved in resource acquisition. Study of the behavioural phenotypes of Angelman and Prader-Willi syndromes could reveal a similar effect of genomic imprinting on social and other behaviours that are consistent with this interpretation.

Haig and Wharton (2003), Brown and Consedine (2004) and Isles and Wilkinson (2000) have suggested that aspects of the phenotypes of Angelman and Prader-Willi syndrome result from lack of expression of alleles responsible for behavioural and cognitive characteristics that are different in function dependent on whether they are beneficial for perpetuating maternally or paternally derived genes. More specifically, Brown and Consedine suggest that some of the behavioural characteristics of Angelman syndrome, in which there is reduced or absent expression of maternally imprinted alleles, function to gain maternal resources when a child is in competition with siblings, thus favouring the paternal line. One behavioural characteristic of Angelman syndrome that has the capacity to command maternal resources is excessive laughing and smiling and Brown and Consedine draw on emotion signalling theory to explain how smiling in Angelman syndrome would have the effect of evoking high levels of social contact.

Few studies have examined smiling and laughing in Angelman syndrome using psychometrically robust assessments and adequate control groups. However, in a review of 64 case and cohort studies of 842 cases, 56 studies identified an elevated predisposition for laughing and smiling and high levels of sociability (Horsler & Oliver, 2006a). This literature is inconsistent in interpreting the cause of the laughing and smiling, with the majority of authors suggesting environmental factors are not influential (e.g., Dooley, Berg, Pakula, & MacGregor, 1981; Williams & Frias, 1982) and a small number of papers noting a lowered threshold for laughing and smiling in social and other settings (e.g., Yamada & Volpe, 1990). In the first study to experimentally investigate the effect of social contact on laughing and smiling in children with Angelman syndrome, Oliver, Demetriades, and Hall (2002) demonstrated that three children with the syndrome laughed and smiled more when social interaction was ongoing than in control conditions. Horsler and Oliver (2006b) extended this finding in a study of thirteen children with a confirmed deletion and also showed that smiling, touch, eye contact and speech from adults were important in eliciting smiling and laughing. In combination, these studies show that smiling is elicited by social contact, demonstrating the behaviour is socially functional, and this is consistent with the hypothesis that the behaviours might increase social resources.

To evaluate Brown and Consedine's hypothesis it is necessary to examine whether children with Angelman syndrome access higher levels of social contact from mothers than siblings and whether episodes of laughing and smiling are influential in accessing this social contact. However, this comparison will be confounded by differences in age and degree of intellectual disability between the children with Angelman syndrome and typically developing siblings. A less direct, but better controlled, comparison is whether children with Angelman syndrome access more social resources than age, gender and degree of intellectual disability comparable children in a socially competitive setting. This comparison can be made in school settings.

There are three aims to this study. First, we compare the percentage of time children with Angelman syndrome engage in smiling and laughing and social approaches toward adults to that shown by children of similar age, degree of intellectual disability and gender (comparison group) in naturally occurring settings which demonstrably differ in the amount of social contact available from an adult. In line with previous studies, we predict that the percentage of time children with Angelman syndrome spend laughing and smiling will both be higher than the comparison group and vary according to the amount of social contact available. Second, we examine whether laughing and smiling in children with Angelman syndrome evoke social contact from adults in a socially competitive environment and are more effective in accessing social resources than when shown by children in the comparison group. Third, we examine child initiated contact to establish whether and how this behaviour might be related to laughing and smiling.

Method

Participants

Thirteen children with Angelman Syndrome (AS; eight male, five female) were recruited via the Angelman Syndrome Support, Education and Research Trust. All had a confirmed deletion of 15q11-13. The mean age was 8 years 2 months (range: 4–17 yrs). A comparison group of ten children with intellectual disability of heterogeneous cause (six male, four female) were selected by matching for age, gender and level of intellectual disability. For seven of these participants aetiology of intellectual disability was unknown; the other three participants had diagnoses of Down syndrome, cerebral palsy and Prader-Willi syndrome. The mean age of the comparison group was 9 years 8 months (range: 5–17 yrs). The mean mental age equivalence, as measured by the Vineland Adaptive Behavior Scale (Sparrow et al., 1984), was 18.85 months (range 11–27 months) for the AS group and 23.50 months (range: 10–42 months) for the comparison group. Participants in the comparison group scored below the cut-off for autism on the Childhood Autism Rating Scale (range 17.50 to 29.50; mean = 25.40, SD = 4.50). Whilst it would have been preferable to have equal numbers in the groups, appropriate matching within comparable environments precluded this.

Procedure

Each participant was observed for approximately four hours in school settings. Observations were videotaped and the frequency and/or duration of target behaviours recorded using OBSWIN data collection software (Martin, Oliver, & Hall, 2001). This software records streams of observational variables simultaneously in real time. Eight variables were recorded. These were the behaviours of both the participant and the school staff and the setting. Codes for settings and behaviours were operationally defined and agreed prior to observation.

Participant variables were *child smile (*stretching of the lips and upturning of the corners of the mouth, frequently followed by parting of lips and viewing of teeth) and *child initiated contact* (reaching for, touching an adult). Staff variables were *adult smile*, *eye contact* (gaze of both adult and child directed at each other's eyes simultaneously) and *adult attention* (talking to, touching child). Three settings were: *one to one attention* (direct attention from adult to child with no other child present), *shared attention* (group setting with adult engaging in activity with two or more children) and *low attention* (no structured activity, free to play without instruction or expectation from staff). Inter-observer reliability was calculated for approximately 20% of total observations across participants. Agreement was calculated based on 10 s intervals for the presence of each variable. Kappa was at least .7 for each variable (mean = .91; range .77 to .96).

Observational data analysis

To examine sequential relationships between child and adult behaviours, two lag analysis procedures were employed. In the first the sequence of the onset of child and adult smiling was examined. Lags were examined for each participant in 10 s intervals prior to and subsequent to the *onset* of child smile and the conditional probability of the *onset* of adult behaviours (smiling, eye contact and adult attention) was calculated within these intervals. In using the *onset* of a child smile and lagging to the *onset* of an adult smile it is possible to isolate adult smiles that occur after the child has smiled as opposed to capturing those that continue past the time that the child starts to smile. Additionally, by *restricting* the analyses up to the onset of the next child smile it is possible to ensure that the adult smiling can be attributed to the child smile used as the criterion and not to subsequent child smiling. In this way we can determine whether it is the child smiling that triggers the adult smiling or vice versa. In the second analysis the *effect* of child smiling on adult behaviour was examined using six 10 s intervals subsequent to child smiling and lagging from the *onset* of a child smile to the *presence* of an adult behaviour.

Paired *t*-tests were used to determine whether the conditional probability of the onset of adult behaviour differed from the unconditional probability at each time point within groups. In this way the effect of a given condition (child smiling) on adult behaviour could be evaluated between the groups. In these analyses the criterion was the *onset* of a child smiling and the target the *presence* of an adult behaviour. Further lag analyses examined the sequential relationship between child smiling and child initiated contact behaviours (reach for, touch adult) using 10 s intervals for sixty seconds prior to the onset of child smiling. In this analysis the criterion was the *onset* of child smiling and the target was the *presence* of child initiated contact. These latter analyses were unrestricted, that is, further child smiling was allowed into the analysis to allow for the possibility of a reciprocal relationship between child smiling and adult behaviours.

Results

Group comparability

No significant differences were found between groups for age, gender or ability as measured by daily living skills age equivalence score on the Vineland Adaptive Behavior Scale (gender: Fisher's Exact $p = 1.0$; age ($t(21) = .89$, $p = .38$; ability ($t(21) = 1.51$, $p = .16$). The groups are therefore comparable on these variables.

Time spent in settings and levels of adult attention

There were no differences in the amount of time the groups spent in *one to one attention* (AS: 24.7%, C: 18.6%; $t(21) = 1.15$ $p = .26$), *shared attention* (AS: 34.7%, C: 38.7%; $t(21) = .38$, $p = .71$) and *low attention* settings (AS: 41.0%, C: 43.0%; $t(21) = .26$, $p = .80$). A mixed ANOVA was carried out to examine differences in the distribution of adult attention across the three settings. As expected the amount of adult attention received by both groups

varied across the settings (*one to one attention*: 76.8%, *shared attention*: 20.5% and *low attention*: 7.9%) and a main effect for setting was found ($F(2) = 167.97$, $p < .0001$). However, no main effect for group was found ($F(1) = 0.97$, $p = .34$) and there was no interaction effect ($F(2, 42) = 0.62$, $p = .49$). Thus, the groups received a comparable amount of adult attention within each of the settings. Differences in the distribution of eye contact across settings were examined in the same way with a main effect of setting ($F(2,42) = 11.92$, $p = .002$) only, as expected, and no main effect of group ($F(1,21) = 2.10$, $p = .162$). In combination these analyses show that the amount of time spent in each setting, the amount of adult attention received in each setting and the amount of eye contact shared in each setting is comparable across groups. Thus, the children in the two groups were exposed to similar social conditions.

Child smiling and child initiated contact across settings and groups

Figure 1 (left panel) shows the percentage of time children in the two groups spent smiling in each setting. A mixed ANOVA showed significant main effects for group ($F(1,21) = 14.94$, $p = .001$) and setting ($F(2,42) = 8.56$, $p = .001$) in addition to an interaction effect ($F(2,42) = 8.30$, $p = .001$). Post hoc analyses revealed a significant difference in child smiling shown by the groups in all three settings with children in the AS group smiling more. Across settings the AS group smiled more in *one to one attention* than *low attention*, more in *shared attention* than *low attention* but equally in *one to one attention* and *shared attention*. Thus, there is an increased propensity for smiling in AS but also an interaction with social setting whereby the difference in level of smiling seen between the two groups is greater when adult contact is present.

Figure 1 (centre panel) shows the percentage of time the children in the two groups spent initiating contact with adults. A mixed ANOVA revealed a significant main effect of group ($F(1, 21) = 7.60$, $p = .012$) whereby children in the AS group spent more time initiating contact with adults than those in the comparison group. Thus, children with AS show an increased propensity for initiating contact with adults as well as smiling in social settings.

Adult smiling across settings and groups

Figure 1 (right panel) shows that adults spend a greater percentage of time smiling in each of the settings with the AS group than they do with the comparison group. A two-way ANOVA shows this main effect of group to be significant ($F(1,21) = 10.10$, $p = .005$). A main effect of setting was also found ($F(2, 42) = 20.23$, $p < .001$) but this is unsurprising as the amount of time an adult is present (and therefore able to smile at a child) is dictated by the setting. A significant interaction effect was also found ($F(2,42) = 4.69$, $p = .035$). Post hoc analyses showed that adults spent significantly more time smiling in the AS group than they did in the comparison group in the *one to one attention* and *shared attention* settings but not in the *low attention* setting. There were also significant differences in adult smiling for the AS group between *one to one attention* and *low attention* settings, *shared attention* and *low attention* settings and *one to one attention* and *shared attention* settings. As with child smiling, adult smiling occurs for a higher percentage of time for the AS group and this increase is greater in social settings.

The effects of child smiling on adult smiling

Now that we have established that adults and children both smile more in the AS group the question of

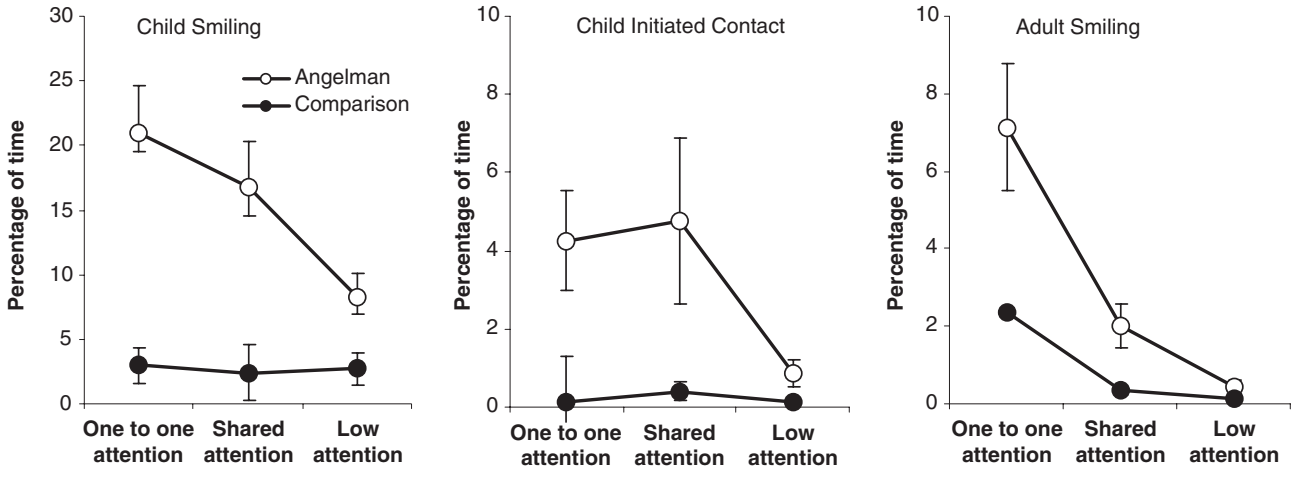

Figure 1 Mean percentage of time (±1 standard error) (a) child smiling, (b) child initiated contact and (c) adult smiling were observed in three settings for the comparison and Angelman syndrome groups

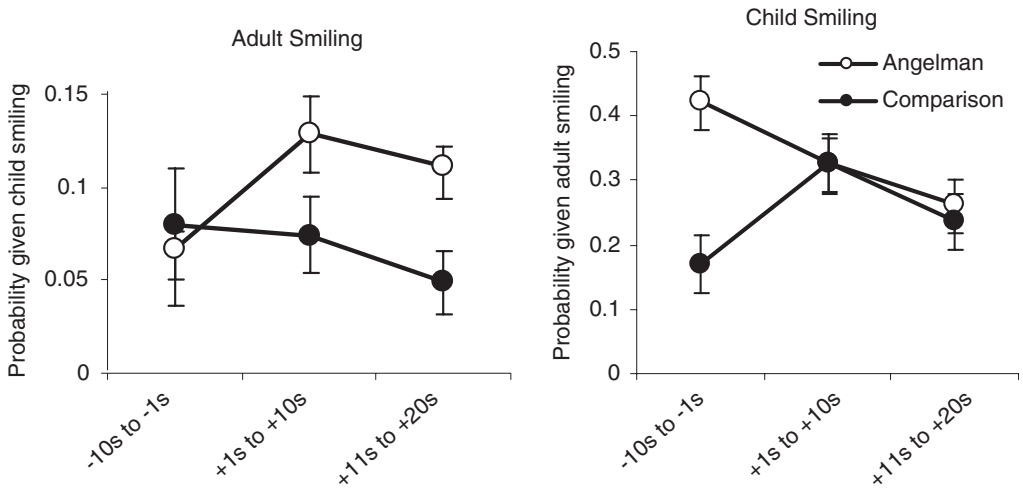

Figure 2 Mean probability (±1 standard error) of the onset of adult smiling prior to and following the onset of child smiling at +1 s (left panel) and the onset of child smiling prior to and following the onset of adult smiling at +1 s (right panel) for the comparison and Angelman syndrome groups

who leads and who follows must be posed. A sequential lag analysis was carried out in order to investigate the sequence of child and adult smiling. Figure 2 (left panel) shows the conditional probability that an adult will start smiling before, during and after a child starts to smile. ANOVA revealed a significant interaction effect ($F(2,42) = 7.97$, $p < .001$) whereby the conditional probability that an adult will smile prior to a child smiling is similar for the two groups but once the child has started to smile the probability that the adult will smile increases in the AS group but not in the comparison group. Thus, it is the child smile which precedes the adult smile in the AS group. In order to confirm this interpretation a second time lag analysis was carried out, this time using onset of the adult smile as the criterion behaviour and lagging to the onset of the child smile (see Figure 2, right panel). Again ANOVA revealed a significant interaction effect ($F(2,42) = 9.40$, $p < .001$) whereby the probability that a child will start smiling prior to the adult starting to smile is higher in the AS group prior to the adult smile whereas in the comparison group the probability of child smiling is lower before the adult smile and increases after the smile. Thus, in the comparison group adult smiling precedes child smiling, whereas in the AS group child smiling precedes adult smiling.

The social consequences of child smiling were examined by lagging forward for six 10 s intervals after a child has smiled and examining whether adult smiling, eye contact and adult attention behaviours (social resources) occur. The conditional probability of an adult behaviour occurring contingent on child smiling was compared to the unconditional probability of that adult behaviour occurring. Analyses were carried out for both the AS and the comparison groups in order to examine differences in the effect that child smiling had on these adult behaviours.

Figure 3 shows the conditional probability of the presence of adult smiling, eye contact and adult attention subsequent to the onset of child smiling. In the comparison group the conditional probability of the three adult behaviours being present does not differ significantly from the unconditional probability following the onset of a child smile. However, in the AS group paired *t*-tests show that the conditional probability of adult smiling, eye contact and adult attention after a child smile is significantly greater than the unconditional probability and this difference is maintained for a period of time. For adult smiling this difference is seen for 50 seconds after the onset on the child smile ($p = .01$ to $.001$), for eye contact the difference is seen for 30 seconds after the child smile ($p = .01$ to $.001$) and for adult attention it is seen for 50 seconds after the child smile ($p = .001$). Thus, adult attention, adult smiling and eye contact are all more likely to occur after a child smile than they are by chance in the AS group, whereas there is no difference between the conditional and unconditional probabilities in the comparison group. Furthermore, this increased probability is sustained after the smile has occurred. These analyses show that child smiling in the AS group solicits more social resources, in terms of adult attention, smiling and eye contact, than the same behaviour in the comparison group.

Child behaviours preceding smiling

In order to determine whether children with AS initiate social contact prior to smiling, a series of time sequential lag analyses were conducted for both groups to calculate the conditional and unconditional probability of child initiated behaviours (reach for or touch adult) at six 10 s intervals prior to the occurrence of a child smiling. Figure 4 shows that there are no significant differences between conditional and unconditional probabilities of child

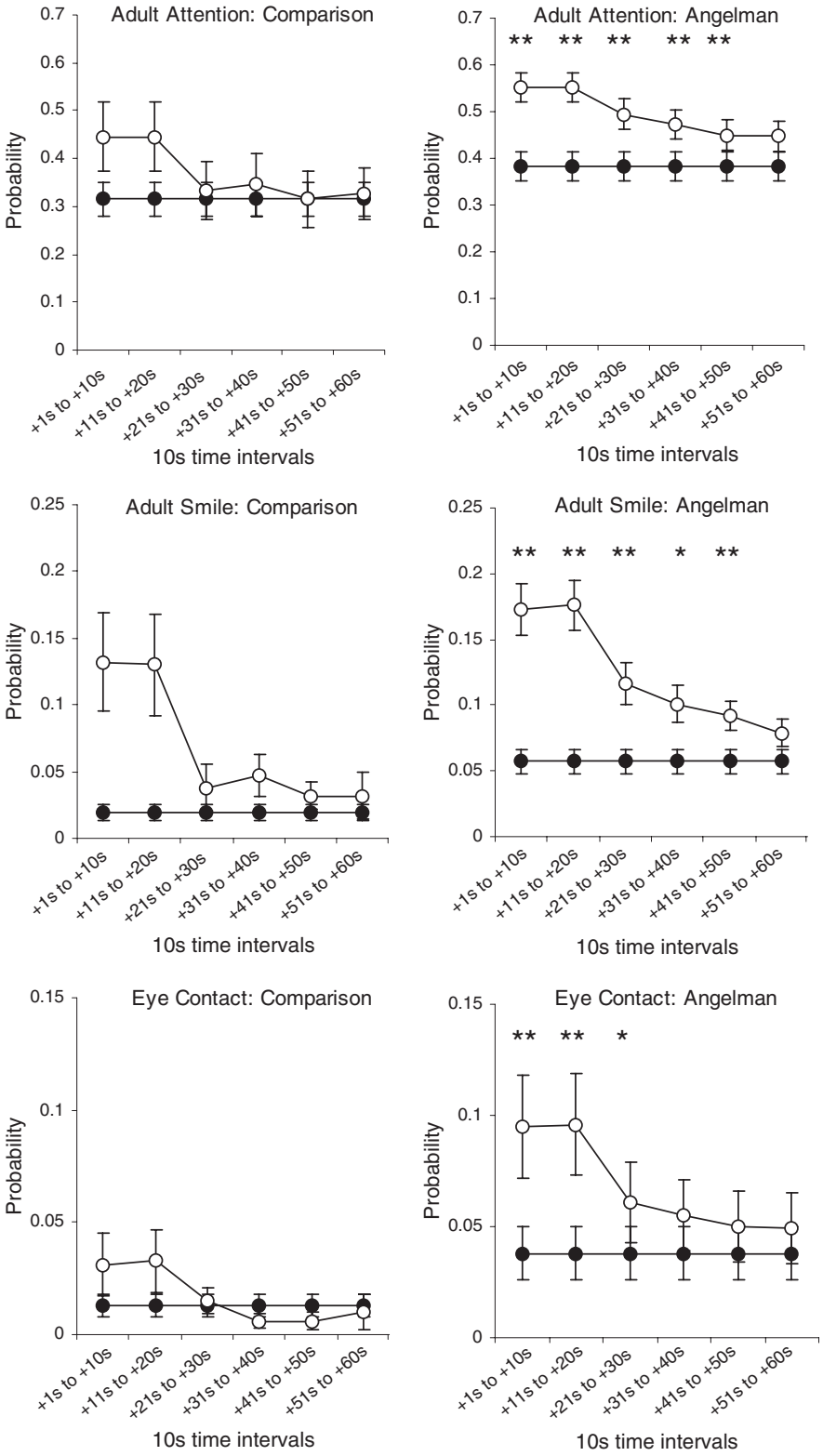

Figure 3 Mean unconditional probability (±1 standard error; filled circles) of the presence of adult smiling, adult attention and eye contact and mean conditional probability (±1 standard error; unfilled circles) of the presence of adult smiling, adult attention and eye contact given the onset of child smiling, for six 10 s intervals following the onset of child smiling for the comparison and Angelman syndrome groups (* = $p < .01$; ** = $p < .001$)

initiated behaviours prior to child smiling for the comparison group. However, in the AS group there is a significant difference between the conditional probability of a child reaching for or touching an adult and the unconditional probability of these behaviours occurring for 30 seconds prior to the onset of the child smile ($p = .001$ to .01). Thus, children in the AS group are significantly more likely to initiate contact with adults prior to smiling at them.

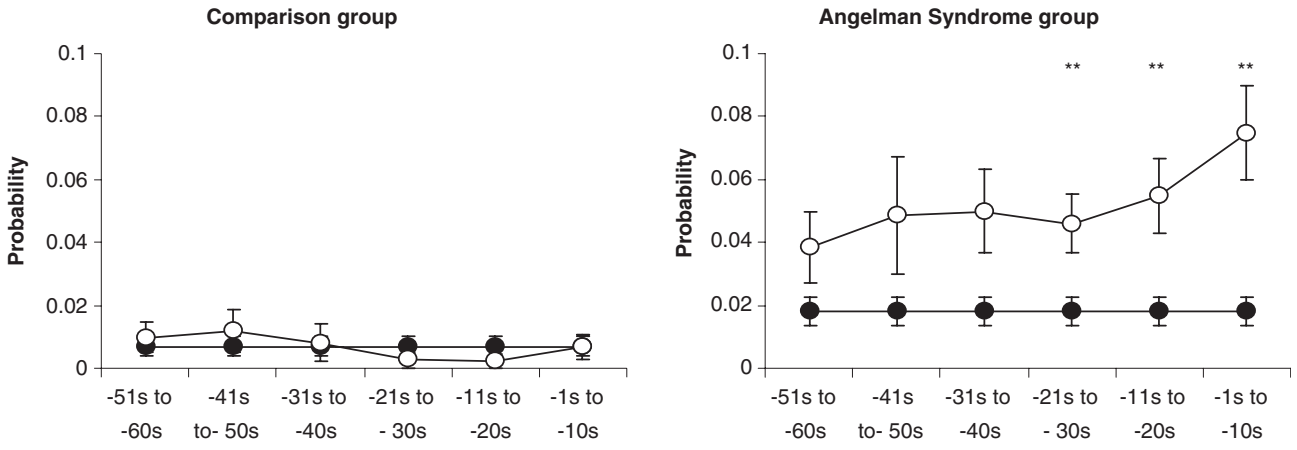

Figure 4 Mean unconditional probability (±1 standard error; filled circles) of the presence of child initiated contact and mean conditional probability (±1 standard error; unfilled circles) of the presence of child initiated contact given the onset of child smiling, for six 10s intervals prior to the onset of child smiling for the comparison and Angelman syndrome groups (* = $p < .01$; ** = $p < .001$)

Discussion

In this study we employed reliable observational codes for behaviour and social settings to compare laughing and smiling by children with Angelman syndrome with that of children of similar age, gender and degree of intellectual disability. The first analyses showed the children with Angelman syndrome smiled more than the comparison group both across settings and when in settings where there was demonstrably more social contact from adults. The latter finding confirms the results of the experimental studies of Oliver et al. (2002) and Horsler and Oliver (2006b) and extends the literature on Angelman syndrome by demonstrating that this effect is phenotypic for Angelman syndrome, as defined by Dykens (1995), when the results for the comparison group are considered. The former finding shows that whilst social contact influences smiling it is not a necessary condition for smiling. This is consistent with previous observations (Dooley, Berg, Pakula, & MacGregor, 1981; Williams & Frias, 1982). Thus, there appears to be both a lowered threshold for smiling when social contact is present and a propensity to smile regardless of whether social contact is ongoing.

Two other findings from these initial analyses were that children with Angelman syndrome initiated more contact with adults than the comparison group and that adults smiled more when with the children with Angelman syndrome. The initiation of social contact by children with Angelman syndrome extends the repertoire of behaviours that might be considered to solicit social resources from adults and confirms, via observational study, previous reports of heightened sociability in Angelman syndrome (Horsler & Oliver, 2006a). The observation that adults smiled more when with the children with Angelman syndrome than children in the comparison group warranted further examination as it may have indicated a confounding variable in analyses (the children with Angelman syndrome might have smiled more because the adults smiled more). However, the analyses of the sequence of the onset of smiling by each player in the dyad showed it was the child with Angelman syndrome who smiled before the adult. Thus, the most parsimonious explanation of the higher level of smiling in the adults is that their smiling was in response to the smiling of the children. This relationship was not evident for the comparison group and this is an interesting evocative gene effect phenotypic of Angelman syndrome. The positive affect experienced by the adult, inferred from the smiling, alludes to the mechanism by which emotion signalling accesses social resources, i.e., the adults finds the interaction rewarding (Wild, Erb, Eyb, Bartels, & Grodd, 2003). This offers support for Brown and Consedine's use of emotion signalling theory as explanatory of the form of behaviours that might access or maintain social resources but identifies the smiling as maintaining rather than initiating social resources.

Following the onset of a smile by a child with Angelman syndrome, the child received significantly higher levels of eye contact and adult attention and adults smiled more. In each case these consequences were significantly more likely after the onset of the smile than by chance and, depending on the behaviour, this effect lasted for between 30 and 50 seconds. For the comparison group none of the adult behaviours were similarly evoked, although a small, but nonsignificant, increase in relevant adult behaviours was evident. These analyses show both that the smiling by children with Angelman syndrome evokes social resources from adults and that their smiles are more effective at doing so than those of comparable children. These results are consistent with the prediction of Brown and Consedine (2004) and show that an effect of smiling in children with Angelman syndrome is to

significantly increase access to social resources in a competitive setting.

It is interesting to consider this result in tandem with the analysis that showed that there was no difference between groups in the overall amount of adult attention received within each setting. At this crude level of analysis the children with Angelman syndrome were not more effective at accessing social resources (amount of adult attention was allowed to naturally vary and can thus be examined in this way). This is inconsistent with Brown and Consedine's prediction. However, the effects on adult behaviour after the smiling were robust. These data show that the effect of smiling is local to the smile and this adds weight to the emotion signalling interpretation; it is smiling specifically that is influential.

It is important to note that children with Angelman syndrome were more likely to have approached an adult in the 30-second period prior to smiling. This relationship was not evident for the comparison group. This is an interesting finding to consider alongside emotion signalling theory. Smiling is only efficient at accessing resources if the 'intended' receiver sees the signal. It is implausible that children with Angelman syndrome plan contact in order to smile and thus access social resources. An alternative explanation is that the children find adult contact highly rewarding and smile as a result of the positive affect they experience. The smile then maintains rather than evokes social contact. This interpretation is consistent with reports of heightened sociability (Horsler & Oliver, 2006a), the observation in this study and previous studies that ongoing social contact evokes smiling (Oliver et al., 2002; Horsler & Oliver, 2006b) and the sequence of children seeking contact, then smiling and subsequently maintaining social contact. The implication of this interpretation is that it is the rewarding property of social contact that is central to the behaviours in the phenotype relevant to genomic conflict rather than smiling, with the latter downstream from initial access of social contact but nevertheless effective in maintaining social resources once accessed.

This interpretation has clinical implications for interventions for children with Angelman syndrome. In an operant learning paradigm, potent social reinforcement from adults might account for the crude aggression, primarily grabbing and hair-pulling (Summers, Allison, Lynch, & Sandler, 1995), observed in Angelman syndrome if adult contact is contingent on these behaviours. It is notable that the specific forms of behaviour, grabbing and hair-pulling are behaviours that would temporarily prolong social interaction. Additionally, if social contact is a potent reward across children with Angelman syndrome, judicious deployment could help with behaviour management programmes. Social contact as a reward would be particularly useful as it is easily administered and, in this case, probably highly resistant to satiation.

One interpretation of these results is that the function of the maternal allele at chromosome 15q11-q13 is to inhibit preference for contact with adults or some social behaviours that access resources from adults. However, as noted by Haig and Wharton (2003), the absence of expression of genes is a substantial deviation from the norm and effects on behaviour should be interpreted cautiously. Additionally, although the results support predictions made by Brown and Consedine with regard to expression of affect in Angelman syndrome, the study is an incomplete appraisal of the kinship theory in this context. We did not test the hypothesis with regard to maternal resources specifically and did not show the opposite relationship for children with Prader-Willi syndrome. This is in part because a comparison of Angelman and Prader-Willi syndromes is confounded by the substantial difference in ability. Further research should examine child–adult interactions in both syndromes and compare predicted outcomes relative to two comparison groups, each matched to a syndrome group. This would overcome the weakness of this study which included one child with Prader-Willi syndrome in the comparison group and did not assess autistic spectrum disorder in the Angelman group. Additionally, the behaviours that kinship theory would predict to differ should be extended to include sleep, feeding and exploration of novel stimuli. Finally, difference in the causal genetic mechanism within both Angelman and Prader-Willi syndromes offers a further extension of this line of research, as does examination of change in behaviours of relevance throughout development.

The findings of this study have a broader relevance to the conceptualisation of the social impairments of autistic spectrum disorder and the potential link between imprinting, gene expression in the developing brain and social behaviour. Autistic spectrum disorder is reported in Angelman syndrome but it is unclear whether this is due to failure on assessments due to the profound intellectual disability, abnormally strong motivation for social contact in the absence of social understanding or the influence on aggregated scores of a subgroup within Angelman syndrome. Further research should focus on modified and appropriate assessment of social behaviour and impairment in Angelman syndrome that might resolve this question. Finally, the association between imprinting and social behaviour in this syndrome indicates the importance of considering functional in addition to structural outcomes for genetic disorders. The abnormally strong social motivation in Angelman syndrome alludes to a role for imprinted genes in the preference for social contact and suggests that consideration of the role of imprinting in autistic spectrum disorders might be fruitful.

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