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Self-injurious Behavior,
Self-Restraint and Compulsive Behaviors in Cornelia de Lange Syndrome

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Running Head: Self-injury in Cornelia De Lange Syndrome

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Abstract

Self-injurious behavior (SIB) has been described as a feature of Cornelia de Lange syndrome (CdLS), with suggestions that the behavior has compulsive properties. It has been argued that individuals who show self-restraint are more likely to show SIB that is compulsive, involuntary and without adaptive function. The aims of this study were to explore the challenging behaviors associated with CdLS, to examine whether self-injury is associated with self-restraint, and to establish whether individuals displaying SIB and self-restraint display more compulsive behaviors than those without SIB and self-restraint. Eighty-eight main caregivers of individuals with CdLS completed questionnaires regarding the behaviors of those they cared for. A significant association was found between SIB and self-restraint and those displaying both of these behaviors displayed significantly more compulsions than those without these behaviors. Findings are interpreted as extending the compulsive behavior theory and highlighting areas for further research.

The Association Between Self-injurious Behavior,
Self-restraint and Compulsive Behaviors in Cornelia de Lange Syndrome

Various behaviors have been reported to be associated with Cornelia de Lange syndrome (CdLS) (Udwin & Dennis, 1995) and self-injurious behavior (SIB) in Cornelia de Lange syndrome has been cited as an example of a behavioral phenotype (Bryson, Sakati, Nyhan & Fish, 1971; Johnson, Ekman, Friesen, Nyhan & Shear, 1976). Other characteristics argued to be associated with CdLS have included individuals not being “talkative, even when they have well-developed vocabularies”, autistic features, aggression (Udwin & Dennis, 1995) and hyperactivity (Greenberg & Coleman, 1973).

A number of case studies have noted SIB in CdLS (Singh & Pulman, 1979; Menolascino, McGee & Swanson, 1982; Dossetor, Couryer & Nicol, 1991; Bay, Mauk, Radcliffe & Kaplan, 1993). However, the behaviour is not always reported (McIntire & Eisen, 1965; Pashayan, Levy & Fraser, 1970; Ptachek, Opitz, Smith, Gerritsen & Waisman, 1963; McArthur & Edwards, 1967; Filippi, 1989). In cohort studies the prevalence of SIB reported varies considerably. Beck (1987) reported “self-mutilation”, presenting as finger-biting, in 16.6 % of a sample of thirty-six. Sarimski (1997) reported rates of self-injurious behavior in 40% of his sample of twenty-seven. Hawley, Jackson and Kurnit (1985) described “behavior management problems including excessive screaming, biting and hitting self and/or others, and frequent tantrums” in 57 % of their sample of sixty-four individuals. Finally, Gualtieri (1990) found 64% of eighty-eight individuals with CdLS showed SIB and Johnson et al., (1976) described seven individuals with CdLS who self-injured in a sample of nine.

A number of observations have been made by authors who have reported SIB in CdLS. Firstly, the preference by individuals who self-injure for imposed physical restraint (Shear et al., 1971; Dossetor et al., 1991). Secondly, although Udwin and Dennis (1995) state that “the self-mutilation in Cornelia de Lange syndrome does not have a compulsive quality” (p. 100), this has been contradicted by authors who have

described the ‘compulsive’ and severe nature of the behavior (Bryson et al., 1971). Shear et al., (1971) reported two cases of SIB in Cornelia de Lange syndrome with a “compulsive” quality and preference for physical restraint, likening it to that observed in Lesch-Nyhan syndrome. These observations and the high prevalence of SIB in this CdLS, have led authors to argue that the SIB found in CdLS may have a biological basis (Bryson et al., 1971; Shear et al., 1971; Johnson et al., 1976).

Although there have been reports of individuals with CdLS becoming distressed when physical restraints are removed (Shear et al., 1971; Dossetor et al., 1991), no study has directly examined the occurrence of self-restraint. A number of individuals with severe mental retardation displaying SIB have been reported to display self-restraint (Powell, Bodfish, Parker, Crawford & Lewis, 1996). Typical topographies include entanglement of limbs in clothing, furniture or body parts (Smith, Iwata, Vollmer & Pace, 1992; Oliver, Hall, Hales, Murphy and Watts, 1998), choosing mechanical restraints (e.g arm splints), and positioning oneself to prevent movement of arms or hands (Powell et al., 1996). Prevalence estimates for self-restraint in those who self-injure range from 10-50% (Smith et al., 1992).

Self-restraint is one phenomenon cited by King (1993) as supportive of a Compulsive Behavior hypothesis of SIB. His theory has been based primarily on two observations. Firstly, authors have commented on the ‘compulsive’ nature of some types of self-injurious behavior that appear unrelated to environmental contingencies and seem to be “involuntary” (Dizman & Cheatham, 1970). Secondly, studies have reported individuals attempting to refrain from self-injurious behavior, (Buzas, Ayllon & Collins, 1981; Christie, Bay, Kaufman, Bakay, Borden & Nyhan, 1982).

King (1993) argues that self-restraint functions as an attempt to resist compulsive SIB, which is similar to individuals with Obsessive-Compulsive Disorder (OCD) who attempt to refrain from their compulsions. King’s theory asserts that for a subgroup of self-injurers, self-injury is an involuntary compulsive behavior, without adaptive function, related to cerebral damage and occurring when anxiety is the setting event. He suggests that any situation that precipitates anxiety should increase the likelihood of SIB occurring, and it is this anxiety state that the individual seeks to terminate.

King's theory contrasts with the majority of hypotheses regarding the causes of self-injurious behavior, where environmental determinants have been emphasised. Operant theories have argued that SIB is maintained by positive reinforcement (eg., social interaction) or negative reinforcement (e.g., escape from demands) (Carr, 1977). King's theory postulates that for some individuals the SIB is without adaptive function. This is an important theory to consider in relation to SIB displayed by individuals with CdLS where the behavior has been reported as 'compulsive' and for SIB where no environmental influences are evident. It might be predicted from King's theory that compulsive and self-restraint behaviors should be more common among individuals with CdLS who show SIB.

To date, there have been very few studies of OCD in those with mental retardation. This may be because of the difficulties in the assessment of whether the obsessions are ego-dystonic (perceived to be intrusive, inappropriate and not within the individuals' control) in those with severe or profound mental retardation (Bodfish, Crawford, Powell, Golden & Lewis, 1995). The focus on ego-dystonicity as specified in the DSM-IV (American Psychiatric Association, 1994) has resulted in compulsions being the main focus of the limited research in this area, as compulsive behaviors are more readily observable and identifiable for research purposes (Bodfish & Madison, 1993).

The majority of studies of OCD have not distinguished between individuals with different aetiologies of mental retardation, one notable exception being a study of Prader-Willi syndrome (PWS) by Dykens, Leckman and Cassidy (1996). Although this study did not examine compulsions alongside other problematic behaviors, it is of interest that there are high rates of compulsions reported in PWS, a syndrome in which 95 % of individuals show skin picking (Dykens & Kasari, 1997).

Bodfish et al. (1995) examined the prevalence of compulsions, stereotypic behavior and SIB in adults with severe or profound mental retardation. The occurrence of compulsions was associated with a significantly increased prevalence of stereotypy and self-injury. Powell et al. (1996) expanded on the Bodfish et al. (1995) study and

examined the occurrence of both self-restraint and compulsive behaviors in a sample of individuals who self-injured. They found that 46% engaged in self-restraint and 57% exhibited compulsions. Compulsive behavior occurred significantly more frequently in individuals who engaged in self-restraint and SIB than in individuals who engaged in SIB without self-restraint.

In summary, SIB has been described as a behavioral phenotype of CdLS (Udwin & Dennis, 1995; Bryson et al., 1971; Johnson et al., 1976), based on reports of the high prevalence of SIB. Additionally, the ‘compulsive’ quality of the SIB in CdLS has been reported with suggestions regarding a possible biological basis to the behavior (Bryson et al., 1971; Shear et al., 1971). King (1993) has argued that SIB may be a compulsive, involuntary behavior, without adaptive function, where self-restraint functions as an attempt to resist the compulsive SIB. This has had some empirical support. Compulsions have been associated with stereotypy and self-injury (Bodfish et al., 1995), and compulsive behavior has been reported at higher rates for those engaging in self-restraint and self-injurious behavior (Powell et al., 1996).

Despite the observations of the prevalence and compulsive quality of SIB in CdLS, there has been no study that has examined SIB, compulsions and self-restraint in those with CdLS. The research described in this paper aims to investigate, firstly, the range of challenging behaviors associated with CdLS with a focus on SIB, secondly, whether individuals with CdLS displaying SIB also engage in self-restraint; and thirdly, whether a relationship can be established between SIB, self-restraint and compulsions.

Method

Participants

229 questionnaires were mailed to carers of individuals with CdLS via the CdLS Foundation UK, a parent support group for individuals diagnosed with the syndrome. Included in each pack was a covering letter, participant information and a reply paid envelope. 88 (38.4 %) questionnaires were returned. Informants were asked to indicate when, where and by whom the diagnosis of CdLS was made.

Of the 88 individuals identified with CdLS, 46 (52.3 %) were female and 42 (47.7 %) were male. 80 (90.9 %) individuals were living at home (other than for respite care), and 7 (8 %) were living in residential facilities (1 missing datum). Individuals with CdLS ranged in age from 1 to 38 years (mean = 12.89 years, SD = 8.02). The age at which the individual was diagnosed with CdLS ranged from birth (within 4 hours of the birth in one case) to 15 years old (mean = 1.56, SD = 2.89). 8 (9.1%) individuals were currently being prescribed psychoactive medication. To describe the sample the Wessex Scale was employed (Kushlick, Blunden & Cox, 1973). This scale provides an assessment of social and physical capacity, specifically: vision, hearing, continence, ambulence, speech, self-help and literacy. The data indicated that 48 (56%) individuals had no vision or hearing difficulties, 19 (22%) had hearing difficulties only, 6 (7%) had vision difficulties only and 13 (15%) had both vision and hearing difficulties. 52 (65%) were ambulant, 12 (15%) were partly mobile and 16 (20%) were non-ambulant. On the speech, self-help and literacy (SSL) scale of the Wessex, 6 (7%) were classified as “literate”, 7 (8%) were “able and verbal”, 5 (5.7%) were “able only”, 9 (10.3%) were “verbal only” and 60 (69%) were classified as “neither able, literate nor verbal”.

Measures

The cover sheet for the questionnaires showed the title ‘Research Questionnaire’ to avoid positive bias for self-injury. The first questionnaire was the Wessex Scale and the order of presentation of the questions on each form of challenging behaviour was randomised.

Self-injurious behavior and other challenging behaviors. To identify individuals showing SIB, respondents were asked the following question: “Has the person you care for with Cornelia de Lange syndrome shown self-injurious behavior (e.g., head-banging, head-punching or slapping, removing hair, self-scratching, body hitting, eye-poking or pressing) **in the last month?**”. Carers were also asked whether the individual had shown physical aggression, property destruction and/or stereotypic behavior in the last month. Physical aggression was defined as any punching, pushing,

kicking, pulling hair, throwing objects, or grabbing other's clothing. Property destruction was defined as any tearing or chewing own clothing, tearing newspapers, breaking windows or furniture, slamming doors, or spoiling a meal. Stereotypic behavior was defined as any rocking, twiddling objects, patting or tapping part of the body, constant hand movements, or eye pressing. Test-retest Kappa statistics for administration of these items by interview are .91, .86, .91 and .72 for the questions on SIB, physical aggression, property destruction and stereotypic behavior respectively. Inter-rater reliability Kappa statistics are .71, .62, .72 and .46 respectively (Oliver et al., submitted). No reliability data are currently available for administration of these items by questionnaire.

Self-restraint. The Self-restraint Checklist (Powell et al., 1996) contains descriptions of seven topographies of self-restraint (e.g., “wraps self in own clothing or holds on to own clothing”). Carers were asked to endorse items if the individual had shown the behavior. The scale has an inter-rater reliability of 91% (Powell et al., 1996).

Compulsive behavior. The Compulsive Behavior Checklist (Gedye, 1992) contains twenty five types of compulsions, grouped under five categories: Ordering (e.g. arranges objects in a certain pattern), Completeness/Incompleteness (e.g. insists on closing open doors, open cupboards), Cleaning/Tidiness (e.g. cleans body part excessively), Checking/Touching (e.g. Touches or taps item repeatedly) and Deviant Grooming Compulsions (e.g. Inappropriately cuts hair, eyebrows or pubic hair). Carers were asked to endorse items if the individual had shown the behavior. Psychometric analysis has established good inter-rater reliability (84.8 %), test-retest stability (83.3%) and validity by direct observation (91.4 %) (Bodfish et al., 1995).

Results

SIB and other challenging behavior

Of the sample studied ($N = 88$), 56 (63.6%) individuals had shown SIB in the last month, 50 (56.8%) individuals had shown stereotypic behavior, 47 (53.4%)

individuals had shown property destruction, and 38 (43.2%) individuals had shown aggressive behavior. There was no effect of gender on the prevalence of these behaviors. There was, however, an effect of age on the prevalence of SIB. Those individuals aged 13 and over ($N = 40$) were significantly more likely to show SIB than those individuals aged 12 years and under ($N = 48$), ($\chi^2(1) = 8.49$, $p < 0.005$). There was no effect of age on the prevalence of aggressive behavior, stereotypic behavior or property destruction.

Self-restraint

47 (53.4%) individuals showed at least one form of self-restraint on the Self-restraint Checklist. Table 1 shows the percentage of individuals showing particular forms of self-restraint for each of the 7 categories listed on the Self-Restraint Checklist.

+++ Insert Table 1 here +++

The items “holding onto others” and “holding objects” were the most prevalent forms of self-restraint. The mean number of items endorsed for individuals showing self-restraint was 1.74 ($SD = 0.97$). Of those showing self-restraint, 22 (47.8%) individuals showed one topography of self-restraint and 2 (4.2%) individuals showed more than three forms of self-restraint. Analysis of the effect of age indicated that 65% ($N = 26$) of those aged 13 and over showed self-restraint compared to 44% ($N = 21$) of those aged 12 and under, indicating that self-restraint was more likely in older individuals ($\chi^2(1) = 3.96$, $p < 0.05$). Individuals aged 13 and over also showed a greater number of topographies of self-restraint (mean = 1.23, $SD = 1.27$) than individuals aged 12 and under (mean = 0.69, $SD = 0.93$), ($t(86) = 2.29$, $p < 0.05$). There was no effect of gender on the prevalence of self-restraint or on the number of topographies of self-restraint displayed.

Compulsive behavior

77 (87.5%) individuals showed at least one form of compulsive behavior on the Compulsive Behavior Checklist. Table 1 shows the percentage of individuals showing at least one compulsive behavior in each of the five categories on the Compulsive Behavior Checklist. The table shows that “ordering” and “checking” compulsions

were the most prevalent forms of compulsive behavior. The mean number of categories of compulsions displayed was 2.94 ($SD=1.32$). Of those showing compulsive behaviors 25 (32.5%) individuals showed three categories of compulsive behavior, 15 (19.5%) showed one category, 13 (16.9%) showed four categories, 12 (15.6%) showed two categories and 12 (15.6%) showed five categories. Analysis of the effect of age indicated that individuals in the older age group showed a greater number of compulsive behaviors ($t(85) = 2.32, p < 0.05$) and a greater number of categories of compulsions ($t(86) = 2.12, p < 0.05$) than those individuals in the younger age group. There was no effect of gender on the prevalence of compulsions or on the number of compulsions displayed.

Association between SIB, self-restraint and compulsive behavior.

36 (40.9%) individuals showed both SIB and self-restraint, a statistically significant association ($\chi^2(1) = 7.32, p < 0.01$). Interestingly, 11 (12.5%) individuals showed self-restraint but not self-injury. Figure 1 (upper panel) shows the percentage of cases showing self-restraint cross-classified according to whether or not they showed compulsive behaviors, and whether or not they showed SIB.

+++ Insert Figure 1 here +++

There was a significant association between self-restraint and SIB for those individuals who showed compulsive behaviors, $\chi^2(1) = 6.45, p < 0.05$, but there was no association between self-restraint and SIB for those individuals who did not show compulsive behavior, $p = 1.00$, Fisher's Exact Test. For those individuals who showed compulsive behaviors, a secondary analysis was conducted to determine whether there was an effect of age on the association between SIB and self-restraint. The lower panel of Figure 2 shows that whilst there was no association between self-restraint and SIB in those individuals aged 12 and under ($\chi^2(1) = 0.38, p > 0.05$), there was a significant association between self-restraint and SIB in those individuals aged 13 and over, $p = 0.004$, Fisher's Exact Test.

Given that we had found an association between SIB, self-restraint and compulsive behavior, we examined the association between the number of compulsions shown by an individual and whether or not the individual also showed SIB and/or self-restraint. Figure 2 shows the number of compulsions displayed by an individual for those showing neither SIB nor self-restraint ($N = 21$), those showing either SIB or self-restraint ($N = 30$) and those showing both SIB and self-restraint ($N = 36$).

+++ Insert Figure 2 here +++

One-way analysis of variance indicated that there was a significant difference between the mean number of compulsions shown by individuals in each group, ($F(2, 84) = 11.27, p < 0.0001$). Post hoc analysis using Scheffe's test indicated that those individuals showing both SIB and self-restraint showed significantly more compulsions than those showing either self-restraint or SIB or those showing neither behavior.

Discussion

In this study self-injurious behavior was the most frequently reported challenging behavior, occurring in 63.6 % of the sample. Stereotypic behavior was reported in 56.8% of individuals, disruption and destruction of property or the environment in 53.4 %, and physical aggression in 43.2 % of individuals. Therefore, it is evident that individuals with CdLS experience a wide range of behavioral difficulties, although the behavioral phenotype for CdLS has predominantly described only self-injurious behavior (Bryson et al., 1971; Shear et al., 1971; Johnson et al., 1976). There is clearly the need for future research to examine all the difficult behaviors highlighted in this study, especially as individuals experiencing multiple topographies of challenging behavior may be of particular concern to parents (Hyman & Oliver, 2001).

The prevalence of self-injurious behavior in the last month, reported in this study (63.6%), is similar to that found in a study conducted by Gualtieri (1990). This prevalence is considerably higher than that found in other studies documenting the prevalence of SIB in people with severe mental retardation generally. For example,

prevalence of SIB in a hospitalised population was reported to be 12% (Oliver, Murphy & Corbett, 1987). The study reported here also demonstrated an increase in the prevalence of SIB with age. Although less than 10% of individuals under the age of 10 were displaying SIB in the Oliver et al., (1987) study, in this study, 50% of individuals aged 12 and under were displaying SIB. Whilst this may suggest that SIB is more prevalent and begins earlier in people with CdLS, other factors may have contributed to these higher prevalence rates. For example, the definition of SIB employed in this study did not require tissue damage to have occurred. It is also not possible to conclude that severity is stable with increasing age. The measure employed in this study does not record any parameters of severity.

A significant association was found between self-injury and self-restraint. 64.3 % of individuals displaying SIB also engaged in self-restraint. This is a higher prevalence rate than the 46 % reported by Powell et al. (1996). Additionally, significantly more compulsive behaviors were shown by individuals displaying SIB and self-restraint compared to those without these behaviors. In the sample as a whole, ordering compulsions were the most commonly reported category which is consistent with other studies (Vitiello et al., 1989; Bodfish et al., 1995). However the mean number of compulsions reported in this sample was 5.4 compared to 2.6 found by Bodfish et al. (1995). Further research into the association between self-restraint and SIB is clearly warranted and future studies might benefit from employing measures of self-restraint which go beyond topographical descriptions and include observational methods. Additionally, research using a larger sample to protect statistical validity might consider the association between the behaviors considered in this study and stereotyped behavior, thus extending the findings of Bodfish et al. (1995).

The findings of this study suggest a possible extension of King's (1993) theory by the association found between self-injurious behavior, self-restraint and compulsions. This is the first study to find this association in those with Cornelia de Lange syndrome. King (1993) predicted that compulsive behaviors would be associated with SIB, and this was supported by the higher number of compulsions and categories endorsed in those with SIB and self-restraint, compared to those without these behaviors. However, the fact that there was an association between SIB, self-restraint

and compulsions, is not sufficient evidence to support the hypothesis that SIB is an involuntary, compulsive behavior.

Examination of the prevalence of SIB, self-restraint and compulsions by age group indicated that the prevalence of SIB and self-restraint increased with age. The number of compulsive behaviors displayed by individuals also increased with age. These data underline the importance of developing early interventions targeted at individuals, under 12 years of age, who display SIB.

The findings of this study, however, should be considered in relation to the methodology adopted. Postal questionnaires were used because of the wide geographical dispersion of individuals with CdLS in the UK and because this is a rare syndrome, with a prevalence of approximately 1 in 40,000 births (Ireland, 1996). The response rate of 38.4 % is consistent with other studies using postal questionnaires (Moser & Kalton, 1971; Lebow, 1982). However it must be acknowledged that this is a relatively poor response rate. Additionally, all participants were recruited via the CdLS Foundation and the sample could be unrepresentative of other individuals with CdLS. It could be hypothesised that those caring for an individual with CdLS who also displays challenging behavior may be more likely to join a supportive group in order to access help and information regarding these behaviors. Therefore the levels of challenging behavior in this sample may not reflect the prevalence of challenging behaviors for all individuals with CdLS.

Despite these limitations, data were obtained for 88 individuals with CdLS (88 individuals were also studied by Gualtieri, 1990), exclusively focused on the behavioral aspects of this syndrome. Other large scale studies have predominantly examined the clinical, medical and developmental features of CdLS (See Jackson, Kline, Barr & Koch, 1993; Kline, Barr & Jackson, 1993; Kline, Stanley, Belevich, Brodsky, Barr & Jackson, 1993; Ireland, 1996).

The findings of this study have a number of important research and clinical implications. King (1993) has suggested that the self-injury for some individuals is a compulsive involuntary behavior associated with cerebral damage. However, the exact

nature of the possible location of this damage is unknown, as is the precise nature of possible neurotransmitter involvement, although it has been suggested that the serotonergic and dopaminergic systems may be involved (King, 1993). Thus, SIB is argued to be a compulsive behavior, or repetitive activity, directly resulting from a functional biological disturbance. Alternatively, it has been hypothesized that there may be a more complex pathway, where SIB occurring in younger children is environmentally reinforced, which introduces the repetitive behavior into the individual's behavioral repertoire. There may then be a biological threshold over a period of time where the SIB may activate neurochemical systems and become compulsive (Oliver, 1995). Longitudinal studies are needed to establish empirical support for this hypothesis. If this hypothesis is supported, it would indicate the importance of developing early interventions to manage SIB before it becomes established in the behavioral repertoire and accesses biological mechanisms.

In conclusion, this study found a high prevalence of SIB, physical aggression and destructive behaviors in CdLS. However, because there were no matched controls conclusions can only be drawn about this sample, rather than in CdLS generally. The most interesting and previously unreported finding was the association between SIB, self-restraint and compulsions. Individuals with SIB and self-restraint displayed significantly more compulsions than individuals without both of these behaviors. This finding is not sufficient in itself to argue that SIB is a compulsive behavior. However, the prevalence of self-restraint may indicate that there is a compulsive quality to the behavior, but further studies are required to explore these findings. Given the fact that not all individuals with CdLS display self-injury, compulsions and self-restraint, it is most likely that the development of these behaviors is multifactorial. Prospective studies may be able to isolate the factors that are predictive of this association occurring. The benefits of the compulsive behavior hypothesis of self-injury is that it provides additional elements which contribute to understanding certain types of self-injury, and this may inform more effective clinical interventions.

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Figure Captions

Figure 1. Upper panel: Percentage of individuals showing self-restraint for those showing compulsive behavior and/or SIB. Lower panel: Percentage of individuals showing self-restraint for those individuals who showed SIB and/or compulsive behaviors cross-classified by age group.

Figure 2. The number of compulsions displayed by individuals showing neither SIB nor self-restraint, those showing SIB or self-restraint and those showing both SIB and self-restraint.

Table 1. Percentage of individuals showing particular forms of self-restraint and compulsive behaviors.

Behavior	%
Self-restraint (N = 47)	
Holds onto others/holds onto others clothing.	42.6
Holds or squeezes objects	36.2
Wraps self in own clothing or holds onto own clothing	29.8
Chooses to wear particular item of clothing.	29.8
Holds hands together, holds onto self.	21.3
Positions self to restrain	14.9
Chooses mechanical restraint	0.0
Compulsions (N =77)	
Ordering	71.4
Checking/Touching	63.6
Completeness/Incompleteness	58.4
Cleaning/Tidiness	50.6
Grooming	49.4



