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ORIGINAL CONTRIBUTION

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Behavioural characteristics and autistic features in individuals with Cohen Syndrome

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Abstract Diagnostic criteria for Cohen Syndrome are based largely on physical characteristics, and systematic information about behaviour and social functioning is limited. Typically, individuals with this condition are described as being very sociable and as showing low rates of pathology. However, recent studies have indicated that behavioural difficulties may occur more frequently than previously suggested and that autistic features may be relatively common. The present investigation of 45 individuals with Cohen Syndrome (age 4–48 years) found that, although 57 % of the sample were reported as showing some behavioural disturbance, problems related mainly to anxiety and social interactions; marked anti-social behaviours were rare. Twenty-two individuals met criteria for autism on standardised diagnostic assessments, although the "autistic profile" was somewhat atypical. The implications of these findings for our understanding of Cohen Syndrome are discussed.

Key words Cohen Syndrome – autistic disorder – behavioural characteristics

Introduction

Cohen Syndrome is a rare autosomal recessive disorder, first described in 1973 [3]. A gene (COH1; chromosome 8q22) [14] has recently been identified, although there are indications that there may be considerable genetic heterogeneity [14]. Estimated prevalence is 1:105,000 [13]. To date over 100 reports of individuals with Cohen Syndrome have been published, but most are single case or small group studies and relatively few involve large samples. Moreover, although the various physical abnormalities associated with Cohen Syndrome have been widely documented, there is still a lack of consistency in diagnosis. The following physical anomalies are generally noted as being important diagnostic indicators [1, 6, 11–13]:

(1) Ophthalmologic abnormalities (including retinochoroidal dystrophy, retinal mottling, retinal pigmentosa, myopia)
(2) Microcephaly
(3) Haematological abnormalities (neutropenia)
(4) Facial characteristics (including short philtrum, high arched palate, thick hair and eyebrows, low hairline, protruding/crowded teeth, down-slanting palpebral fissures, prominent nose)
(5) Hand and feet abnormalities (slender, tapered, hyper-extensible)
(6) Truncal obesity
(7) Other physical features (hypotonia, scoliosis, short stature)

Intellectual impairment is considered to be an essential criterion by some groups of researchers [12]. Many descriptive accounts also note low rates of behavioural problems and comment on the sociability and 'cheerful disposition' of individuals with Cohen Syndrome [11, 12, 18, 19].

However, there is disagreement even about the core physical features. Cohen (personal communication), for

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example, notes that retinal pigmentosa is not an appropriate diagnostic symptom as this generally leads to complete blindness, which is not typical of most people with Cohen Syndrome. Moreover, features such as hypotonia, scoliosis and short stature are far from specific. There is even less agreement concerning other characteristics. Thus, a recent study of cognitive and adaptive skills [10] indicated that some individuals may have an IQ in the normal range, and although some research [13] has reported low levels of maladaptive behaviour and high levels of self-direction, responsibility and socialisation, there are also accounts of greater behavioural disturbance [2, 22]. Chandler and colleagues [2], for example, found that the average score for maladaptive behaviour on the Vineland Scales [21] fell within the "intermediate" range and some individuals scored within the "significant" range. In addition, despite anecdotal reports of good social abilities, autistic-type symptoms have been noted in a number of studies. Fryns et al. [7] reported autistic behaviour patterns in four patients and in a postal survey of 33 children and young adults with Cohen Syndrome [9] Howlin found that over half the sample had problems in social understanding, communication and ritualistic and stereotyped behaviours. Kivitie-Kallio and her colleagues [11, 12] noted that "inappropriate interpersonal manners, stereotyped behaviour and odd mannerisms were not uncommon". One of their cases had also shown autistic behaviour as an infant, although this had improved after the age of 3 years. In the Chandler et al. study [2] of 27 patients, 74% exhibited stereotyped behaviours, such as spinning, and five cases (18%) were observed to show autistic features (communication and social abnormalities and ritualistic and obsessive behaviour).

In view of the current confusion about behavioural characteristics, the aim of the present study was systematically to assess patterns of social and behavioural functioning in individuals diagnosed with Cohen Syndrome and to explore the possible association with autistic features. Such information was considered important because, in the absence of a reliable genetic test, and persisting inconsistencies concerning the core physical characteristics, identification of common behavioural characteristics could assist in refining clinical diagnostic procedures. Furthermore, systematic information about the particular behavioural difficulties associated with Cohen Syndrome is essential for families, as such knowledge has major implications for intervention and educational provision.

Subjects and methods

Recruitment

Screening questionnaires and a letter inviting participation in the study were distributed to the families of all 98 individuals registered with the UK-based Cohen Syndrome Support Group. This also has members from Europe, the US, Australia and New Zealand. The families of 76 individuals responded, but for practical reasons only those living in the UK, Ireland or Denmark (n = 64) could be visited for detailed assessments. The families of 51 individuals agreed to participate in the full study. (There were no significant differences in terms of variables such as sex, age, level of schooling, self-help, and language skills between participants whose families did or did not take part in the full study.)

Diagnostic ascertainment

As noted above, there is currently no widely available genetic test for Cohen Syndrome and diagnosis is made primarily on the basis of physical characteristics. Ratings of diagnostic certainty in the present study were based on two measures – the reported certainty of the diagnosing professionals' opinion and the presence of the seven physical abnormalities typically associated with Cohen Syndrome [11, 12]. (See list of items 1–7 above.)

A diagnostic rating of 'Definite' was assigned to those individuals whose diagnosis was described as 'Certain' and who had at least five of the seven principal characteristics. A rating of 'Probable' was assigned to those individuals whose diagnosis was described as 'Certain' or 'Most Likely' and who had at least four of the seven characteristics. A rating of 'Possible' was assigned to those who had fewer than four characteristic features, regardless of clinical rating. Forty-five individuals were rated as having a definite (n = 20) or highly probable (n = 25) diagnosis of Cohen Syndrome; the six cases who were rated as "Possible" were excluded from the analysis. The mean number of Cohen Syndrome features in the "Definite" group was 6.4 (sd = 0.49); in the "Probable" group, it was 4.8 (sd = 0.75). The following analysis of the findings takes account of these differences in diagnostic certainty.

Participants

The average age of the 45 participants was 16.5 years (sd 9.3 years; range 4.8–48.9 years); just over half (26) were female. Two individuals had been raised by foster parents. Two adults were living independently, three were in residential day centres or group homes and two were

still at college; the remainder lived at home. All but one family was Caucasian and the main language in all but two households was English (these were two Danish families who were assessed and interviewed by JK in Danish). Most parents (n = 37) were married or cohabiting, the remainder were single, divorced or widowed. Six families each had two children with Cohen Syndrome.

The average age at which participants had been diagnosed as having Cohen Syndrome was 9.9 years (sd 6.6, range 1.6–36 years). Thirty-six cases had been diagnosed with Cohen Syndrome by a geneticist; seven by a paediatrician and two by an ophthalmologist. Before the diagnosis of Cohen Syndrome had been confirmed, 25 participants had been given alternative diagnoses: these included autism (five cases, four of whom were still considered to be autistic) and a variety of unconfirmed genetic conditions such as Prader-Willi Syndrome (four cases, all subsequently discounted), Schwachman syndrome (n = 2), Bardet-Biedl/Lawrence Moon syndrome (n = 4), unspecified chromosomal abnormalities (n = 2), and single case diagnoses of Rett syndrome, "Dwarf syndrome", Fragile X, Battens disease and Diamond Schwartz syndrome. Others had received somewhat non-specific diagnoses such as mental retardation, hypotonia, dyspraxia, hyperthyroidism and cerebral palsy.

Intellectual and language ability

Non-verbal IQ levels were assessed using the Wechsler Abbreviated Scale of Intelligence (WASI) [23] or, for individuals failing to score on this, the Raven's Coloured Progressive Matrices [20]. If both these proved too difficult, the Vineland Adaptive Behavior Scales (Survey form) [21] were used to derive an overall level of functioning. Comprehension of language was assessed primarily by means of the British Picture Vocabulary Scale-II (BPVS-II) [5] and expressive language by the Expressive One-Word Picture Vocabulary Test-Revised (EOWPVT-R) [8]. For those individuals who could not score on either of these tests, scores on the Vineland subdomains of expressive and receptive language were used to provide estimates of verbal ability.

The average non-verbal IQ or IQ equivalent was 51.8 (sd 22.2; range 20–106). Nineteen individuals obtained a non-verbal IQ within the moderate to severe range (IQ < 50); 17 scored within the mildly impaired range (non-verbal IQ 50–69); 9 had non-verbal IQ scores over 70. The mean receptive language age equivalent was 6.3 years (sd 4.4; range 8 months to 17 years); the mean expressive language age was 5.6 years (sd 4.2; range 8 months to 19 years). (For full details of intellectual and linguistic ability, and levels of adaptive behaviour etc., see Karpf et al. [10]).

Behavioural assessments

Behavioural difficulties were assessed using the parent-based version of the Developmental Behaviour Checklist (DBC-P) [4]. This assesses problems in five main areas: (1) Disruptive and Antisocial Behaviour; (2) Self-Absorbed Behaviour; (3) Communication Disturbance; (4) Anxiety, and (5) Social Relating. The Maladaptive section of the Vineland Adaptive Behavior Scales [21] was used as an additional measure of behavioural difficulty.

The presence of autistic type behaviours was assessed using the Autism Diagnostic Observation Schedule (ADOS) [16, 17]. The ADOS is a semi-structured, standardised observational measure of communication, social interaction and play/imagination. The diagnostic algorithm is based on scores on the Communication and Social Scales, with a cut-off score of 7–8 (depending on the module used) indicating autistic spectrum disorder (ASD) and a score of 12 indicating autism. Caregivers were also interviewed on the Autism Diagnostic Interview-Revised (ADI-R [15]), which focuses on abnormalities of communication, social interactions, and ritualistic and stereotyped behaviours. Cut-off scores for abnormality are: Reciprocal social interaction, 10+; Communication, 7+ for non-speaking individuals, 8+ for verbal individuals; Ritualistic and stereotyped behaviours, 3+. The diagnostic algorithm is based on scores in each of these three domains and symptoms should be evident prior to the age of 3 years. The ADI-R also contains some general questions about early development.

All assessments were conducted by JK. The ADI-R and ADOS require specialist training and, in addition to being fully trained in their administration, JK attended group consensus meetings to ensure reliability was maintained in scoring the ADOS. ADI-R ratings were checked with PH.

The ADI-R interview and Vineland scales were completed for the entire sample; the ADOS was conducted on 43 individuals. Two people could not attempt the ADOS because of blindness, but although another 20 had some visual loss, their parents/caretaker did not consider that this would significantly affect their performance on the modules used. The DBC-P was completed for 44 people (one mother was ill at the time of this interview).

Results

(Note: tests of significance were two-tailed, and a p value of < 0.01 was set in determining significance. Parametric tests were used as far as possible where the data allowed. In comparative tests, the appropriate adjustments were made if the variances were not equal.)

In reporting the data, any differences between the "Definite" and "Probable" diagnostic sub-groups are explored, as are differences in scores related to sex, age group (classified as 4–12 years; 13–18 years and 19 years+), and IQ group (classified as non-verbal IQ 70+; 50–69, and < 50).

Behavioural disturbance

Mean scores on the DBC-P and Vineland Maladaptive scale are presented in Table 1. On the DBC-P, the mean score was 50.2; sd 22.2 (47.3 sd 22.8 in the "Definite" and 52.6 sd 21.9 in the "Probable" diagnostic groups; p of difference = 0.44). Twenty five individuals (11 in "Definite" and 14 in "Probable") scored above the clinical cut-off score of 47. However, when scores on the individual subscales were compared with norms for other groups of individuals with mild-moderate intellectual impairments [21], the present sample had much lower scores for Disruptive/Antisocial Behaviour, and rather higher scores for Anxiety and Social Relating. There was no difference in total DBC-P scores, or scores on any of the sub-scales, between the sub-groups of individuals with a "Definite" or "Probable" diagnosis of Cohen Syndrome (p values ≥0.09 for each comparison). Scores of males and females were also similar (p values ≥0.2 for each comparison). However, there was an effect of IQ for both the Self-Absorbed scale (df 2,41 F = 6.12, p = 0.005) and the Social Relating scale (df 2,41, F = 5.94, p = 0.005), with scores being significantly higher in the moderate to severely intellectually impaired group than in the IQ 70+ group (Self-Absorbed scale p = 0.01; Social Relating scale p = 0.005). There was also an effect of age for the Self-Absorbed scale (df 2,41, F = 5.77, p = 0.006), with individuals in the 19+ age group having significantly lower scores than children in the 4–12 year age group (p = 0.01).

The DBC-P score was highly correlated with the Vineland Maladaptive Behaviour Score (r = 0.83) with the latter indicating "Intermediate" levels of disturbance in 28 cases and a further 12 individuals showing "Significant" levels of disturbed behaviour (see Table 1). The mean total score on the Vineland Maladaptive scales (1 + 2) was 17, which falls within the intermediate range for individuals with mental retardation. Five individuals obtained scores within the non-disturbed range (0–8); 28 were in the intermediate range (9–20) and 12 scored within the significantly disturbed range (21+). Again the scores of individuals with a "Definite" or "Probable" diagnosis of Cohen Syndrome did not differ (Definite = 16.4; sd 7.4; "Probable" 18.2; sd 8.2; p = 0.45). There was no relationship between Maladaptive scores and sex, age, or IQ level (p values ≥0.20 for each comparison). The most commonly occurring problems on the Vineland (i. e. those reported for at least 50 % of the sample) were poor concentration/attention difficulties (42 individuals); poor eye contact (n = 33); sullenness/stubbornness (n = 31); eating disturbances (n = 28); anxiety problems (n = 27); peculiar preoccupations (n = 26); temper tantrums (n = 25); and lability of mood (n = 22). In contrast, few individuals were reported as having definite problems in areas such as truancy (n = 0) or running away (n = 1); teasing/bullying (n = 2); physical aggression (n = 3); lying/cheating/stealing (n = 3); swearing (n = 3); over-activity (n = 3); negativity/defiance (n = 5); lack of consideration for others (n = 5); or avoiding school or work (n = 5).

Parents were also questioned about two other areas in which individuals with Cohen Syndrome are typically reported as having problems – hyperacusis and eating difficulties. Twenty-nine individuals were reported as

Table 1 Scores on Developmental Behaviour Checklist (DBC-P) and Vineland Maladaptive Behavior Scale

DBC-P (N = 44)	Mean	SD (range)		
Total	50.18	22.21 (6–91)	(Clinical cut off = 47)	N at/above cut-off = 25
			Mean for mild/moderate IQ groups[a]	N at/above 70%ile[a]
Disruptive behaviour	10.32	7.61 (0–28)	15.76	14
Self-absorbed	10.68	7.50 (0–32)	11.27	13
Communication disturbance	4.23	3.52 (0–14)	5.97	16
Anxiety	7.93	3.95 (1–19)	4.28	19
Social relating	5.95	3.08 (0–13)	4.21	21
Vineland Maladaptive Behavior Parts 1 and 2 Total (N = 45)	17.44	7.88 (2–32)	"Intermediate" range = 9–20[b] "Significant" range = 21+	N "Intermediate" 28 "Significant" 12

[a] Percentile and cut-off data based on DBC-P norms for individuals with mild-moderate retardation
[b] Ratings of intermediate/significant disturbance based on Vineland figures for mentally retarded individuals (18 years+) in non-residential facilities. The manual does not provide data for younger mentally retarded children

being unusually sensitive to noise and for three this could be so extreme as to interfere significantly with family or household routines. Over three-quarters of the group (35 individuals) had marked eating problems as infants, often apparently due to congenital anomalies of the larynx (two individuals had a tracheostomy for the first few years of their life). With age, however, the most frequent problems related to overeating. Almost two-thirds of the group (n = 27) tended to eat excessively and 36 were reported to have weight problems. Eating disturbance was also reported on the Vineland (n = 28).

The presence of autistic features

Algorithm scores on both the Autism Diagnostic Interview-Revised (ADI-R) and the Autism Diagnostic Observation Schedule (ADOS) are summarised in Table 2. The average age at which parents first noted that there was a problem in their child's development was 7.12 months (sd 6.87, range 0–30 months). Many parents (n = 10) were concerned within the first month of life and 25 had anxieties by 6 months. Problems related to delayed milestones and feeding difficulties were the cause of most initial concerns (n = 24); problems in social/emotional responsiveness were listed as the first concerns by ten parents and general behavioural difficulties were noted by seven. (The remaining cases had shown a mixture of early symptoms.)

On the ADI-R, 36 individuals reached the cut-off criterion (score of 10+) for problems in social interactions, notably in the areas of developing peer relationships and socio-emotional reciprocity. Many individuals had difficulty sharing and were somewhat disinhibited. Thirty-seven individuals scored at or above the cut-off for problems in communication (score of 7+ for non-speaking individuals and 8+ for verbal individuals), particularly with regards to limited reciprocal conversation, repetitive questioning, and a marked lack of varied spontaneous make-believe or social play. Thirty-nine participants scored at or above the cut-off score of 3+ for ritualistic and stereotyped behaviours such as sorting and lining up objects, unusual sensory interests (e. g. feeling and smelling materials), insistence on following set routines, and stereotyped motor mannerisms. Thirty-four individuals (15 in the "Definite" and 19 in the "Probable" diagnostic groups) met full ADI-R diagnostic criteria for autism in that they scored above the cut-off on all three domains (Social, Communication and Stereotyped behaviours) and met criterion for age of onset.

Of the 43 individuals who were assessed on the ADOS, 34 (17 in both the "Definite" and "Probable" Cohen Syndrome diagnostic groups) met criteria for ASD (score 7 +) and 23 of these individuals (11 "Definite"; 12 "Probable") met criteria for autism (scores 12+).

The proportions meeting autism diagnostic criteria on the ADI-R did not differ in the "Definite" or "Probable" Cohen Syndrome groups and there were no differences on the ADOS in the proportions of either diagnostic group scoring above the ASD or autism cut-offs. For neither instrument was there a relationship between algorithm scores and age or sex. However, there was a relationship between the total ADOS algorithm score and IQ (df 2,42, F = 12.16.036, p < 0.001). Thus, individuals with an IQ in the normal range had significantly lower (i. e. better) ADOS scores than those who were moderately to severely retarded (p < 0.001). The ADOS scores

Table 2 Scores on Autism Diagnostic Interview-Revised (ADI-R) and Autism Diagnostic Observation Schedule (ADOS)

ADI-R (N = 45)	Mean	SD (range)	N at/above autism cut-off	
Total (Cut-off score = 21/22)[a]	34.09	12.93 (6–52)	34	
Impairments in reciprocal social interaction (Cut-off score = 10)	16.58	7.25 (2–28)	36	
Impairments in reciprocal communication (Cut-off score = 7/8)[a]	12.27	5.20 (2–21)	37	
Repetitive and stereotyped behaviours (Cut-off score = 3)	5.24	2.27 (0–10)	39	
ADOS (N = 43)			**N at/above autism cut-off**	**N at/above ASD cut-off**
Total (Cut-off scores: Autism = 12; ASD = 7/8)[b]	10.77	4.76 (1–22)	23	34
Reciprocal Communication (Cut-off scores: Autism = 4/5; ASD = 2/3)[b]	4.05	1.76 (0–8)	34	39
Social interaction (Cut-off scores: Autism = 6/7; ASD = 4)[b]	6.72	3.35 (0–14)	24	37

[a] Cut-off scores for communication on ADI-R vary according to language level
[b] Cut-off scores on ADOS vary according to language level and module used

of those with an IQ in the normal range were also marginally lower than the scores of individuals who were mildly intellectually impaired ($p = 0.016$). On the ADI-R, there was no significant association with IQ (df 2,42; $F = 3.016$; $p = 0.06$), although, again, scores for abnormality were lowest in the 70+ IQ group and highest in the below-50 IQ group.

There was a highly significant correlation between total scores on the ADI-R and ADOS ($n = 43$, $r = 0.59$, $p < 0.001$) and of the 34 individuals who met criteria for autism on the ADI-R, 31 met criteria for an autistic spectrum disorder on the ADOS; 22 of these cases (10 in the "Definite" and 12 in the "Probable" Cohen Syndrome diagnostic groups) also met full ADOS diagnostic criteria for autism. It should also be noted that there was a marked and significant difference between the 22 individuals who met criteria for autism on both measures and those who did not. Total ADI-R and ADOS scores were significantly higher in the "autism" than in the "non-autism" group (p for both comparisons < 0.001). There were also highly significant differences between individuals who met full autistic criteria and those who did not in their scores on the separate ADI-R and ADS domains. In other words the data indicated a clear demarcation between individuals who did or did not show autistic symptoms.

Discussion

Although Cohen Syndrome was first identified over 30 years ago, there continues to be disagreement about the specific combination of physical characteristics required to meet diagnostic criteria. Research into associated behavioural characteristics is even more limited although a general stereotype has tended to emerge of individuals with this condition being cheerful, friendly, good-natured and sociable. Kivitie-Kallio [11] reports that all of the 29 cases in her series had a "cheerful disposition" and she suggests that this, together with "almost total absence of maladaptive behaviour", is one of the principal features of the condition. However, recently, there have been accounts of greater behavioural disturbance [2] and the results of the present study indicated clearly that certain behavioural difficulties were common. On the DBC-P, over half the group showed levels of disturbance that were above the clinical cut-off and on the Vineland Maladaptive scale 27 % of the sample had scores within the "significant" range. Nevertheless, most problems were related to developmental delays (i. e. difficulties of concentration, attention, mood, eating, and temper tantrums). Anti-social activities, such as destructiveness, aggression, bullying, lying, swearing or running away, were rare. Very few individuals were reported to be overactive, and negative and defiant behaviours or lack of consideration for others were also uncommon. It may well be the relative lack of such behaviours that has reinforced the cheerful and sociable stereotype.

Data from the ADI-R and ADOS also indicated relatively high rates of autistic-type symptoms, with 31 individuals meeting criteria for an autistic spectrum disorder on both these instruments and 22 meeting criteria for autism on both. Although Chandler et al. [2] suggest that there is little evidence of an increased prevalence of autism in Cohen Syndrome, their study did not involve any systematic autism-diagnostic assessments. However, they note that the majority of their sample showed stereotyped behaviours.

Although there is an established association between autism and intellectual impairment, the frequency of autistic features found in this study, using standardised diagnostic tools of established validity and reliability, was higher than reported for groups of other intellectually impaired children [15, 16]. Moreover, although autistic symptomatology was somewhat less evident in participants of higher IQ, there were individuals at each IQ level who met criteria for autism. The fact that autism scores were not distributed on an evenly sliding scale throughout the sample, but tended to cluster in the group who exceeded cut-off criteria on both the ADI-R and ADOS, indicates that there may be a sub-group of individuals with Cohen Syndrome who show marked autistic features. Thus, it is recommended that the presence of autistic features should be investigated as part of the diagnostic assessment for this condition. However, it should also be recognised that the "autistic picture" is somewhat atypical. Firstly, there was no evidence of the preponderance of males (typically 4:1) that is found in idiopathic autism. Secondly, psychometric assessment of the same sample, reported in a companion study [10], indicated a rather unusual (for autism) profile of functioning. In particular, that study found no significant difference between receptive and expressive language skills, although the latter are typically better developed in autism. The pattern of Vineland scores in that study was also unusual. Thus, scores on the Socialisation scale were higher than on the other domains, although the items incorporated in this scale (play and leisure, social manners, and interpersonal relationships) are not usually relative areas of strength for people with autism (see Karpf et al. [10] for details). As is the case with a number of other conditions, such as Fragile X, Rett syndrome and Williams syndrome, the frequency of autistic symptoms does seem to be unusually high and recognition of this is clearly important both for clinical practice and research. However, there are also certain atypical features and these, too, must be taken into account.

One major problem with this and other descriptive studies of Cohen Syndrome to date arises from the fact that a gene has only been identified within the last year [14], mutation-detection techniques are not 100 % sen-

sitive [14] and it now seems unlikely that all affected cases do have the COH1 gene (only 32 cases were involved in the Kohlmainen study [14] and there was heterogeneity in the mutations identified). The lack of precise definition of physical features means that, until the genetics of the condition are further clarified and until widespread, reliable genetic testing becomes available, it is important also systematically to collect information on the behavioural characteristics of individuals with a suspected diagnosis of Cohen Syndrome. The present study suggests that communication and social deficits, together with a tendency towards routine and repetitive behaviour, may be additional, important diagnostic indicators in at least some cases. Moreover, as knowledge about the genes involved in Cohen Syndrome improves [14], detailed information on behavioural as well as physical characteristics will be crucial in refining the behavioural phenotype, as it is very possible that cases confirmed with the COH1 gene will show a somewhat different phenotype to those who do not have this gene. If individuals with marked autistic features were found to show a different genetic profile to those who do not exhibit such features, this could also add to existing research on the nature of the genetic abnormalities associated with autism.

In conclusion, since routine genetic testing is not available, and whilst the specific physical features associated with Cohen Syndrome still require more precise definition, it is important for diagnosing clinicians to assess behavioural characteristics as well. It is also essential to move away from the stereotype of people with Cohen Syndrome as having few behavioural or social difficulties. A similar stereotype existed many years ago with regard to Down syndrome and only when more systematic studies of behaviour were conducted did a much more complex picture emerge. The present study suggests that, although anti-social behaviours are rare, symptoms of anxiety are common, and in some individuals autistic-type features are marked. It is very possible that these variations in behavioural characteristics may be related to genetic differences. Awareness of the nature of the behavioural difficulties that may be associated with Cohen Syndrome is important for families seeking advice on how to cope with problems, and in furthering our understanding of this complex condition.

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