The Cerebra Centre for Neurodevelopmental Disorders

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Working wonders for children with brain conditions

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SATB2-associated syndrome: A preliminary analysis

Richards Lab
Birmingham

UNIVERSITY OF BIRMINGHAM

Aston University

WARWICK
MEDICAL SCHOOL

A MESSAGE FROM US

Thank you for taking part in this study. The aim of the study is to describe behaviours shown by individuals with SATB2-associated syndrome. The study is being conducted by researchers at the Cerebra Centre for Neurodevelopmental Disorders at the University of Birmingham, in collaboration with researchers from Aston University and University of Warwick.

You were asked about various behaviours including self-help skills, repetitive behaviours, challenging behaviours, communication skills and health problems shown by the person you care for.

This preliminary analysis is based on questionnaire responses from 69 parents/caregivers. We hope that this analysis is useful to you and the person you care for. Individualised feedback reports based on the responses you provided will also be sent to you once the study is complete.

Thank you for taking part in this study. Research such as this would not be possible without the support of families and caregivers. Many thanks to the SATB2 Gene Foundation for their recruitment efforts and continued support of this study.

If you have any questions or comments, do not hesitate to contact Dr Stacey Bissell on S.L.Bissell@bham.ac.uk

Dr Caroline Richards
University of Birmingham

Dr Stacey Bissell
University of Birmingham

Dr Hayley Crawford
University of Warwick

Dr Jane Waite
Aston University

DEMOGRAPHIC

The information presented in Figure 1 shows the age range of 69 individuals with SATB2-associated syndrome (mean age = 10.20 years; range = 1-36 years), based on caregiver reports completed as part of the online questionnaire. Caregivers provided information relating to 36 females and 33 males with SATB2-associated syndrome.

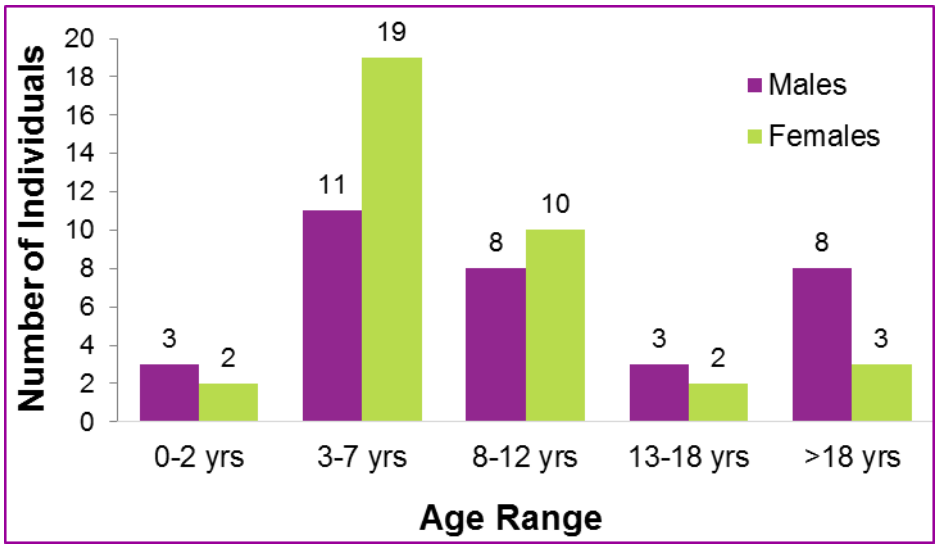

Figure 1. Age distribution of sample according to gender.

As can be seen in Figure 2, 35 families were from the USA, 12 were from the UK, five were from the Netherlands, four were from Canada, four were from Australia and nine families were from mainland Europe.

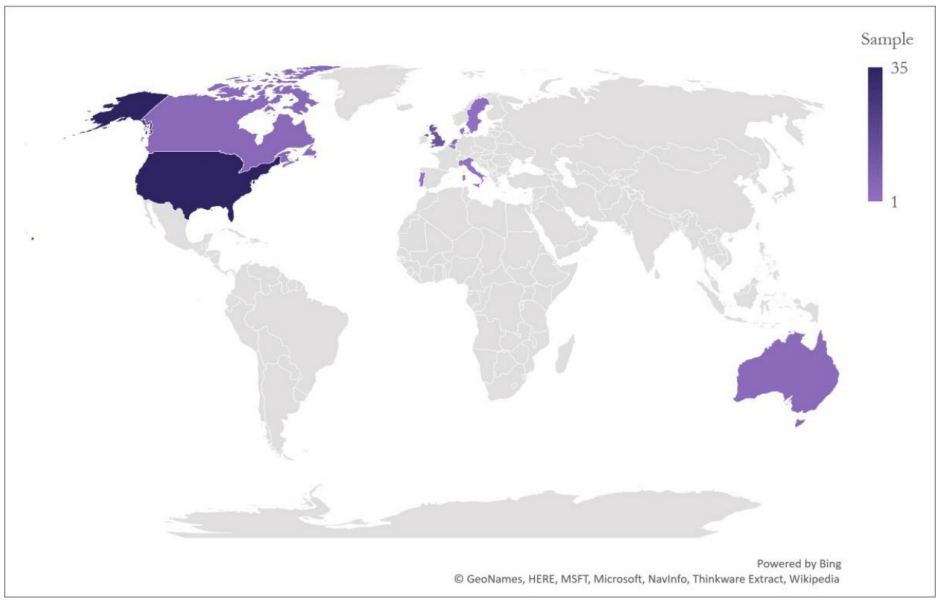

Figure 2. Geographical location of families who participated.

ABILITY AND HEALTH

ABILITY

As outlined in Table 1, most individuals with SATB2-associated syndrome were reported to be mobile with normal vision and hearing. However, speech difficulties were prevalent in the sample. Verbal ability in this study was defined as the ability to communicate using 30 words/signs or more.

Table 1. Physical and communication abilities in SATB2-associated syndrome; N (%).

Fully Mobile	Normal Vision	Normal Hearing	Verbal
63 (91.30%)	57 (82.60%)	65 (94.20%)	22 (31.88%)

As can be seen in Figure 3, most individuals were able to feed themselves independently or with help. In comparison, dressing and washing independently represented areas of difficulty in this sample.

Please note however, the sample included in the present study comprised 16 individuals who were below five years of age. These figures therefore need to be interpreted with caution as developmental factors need to be taken into account.

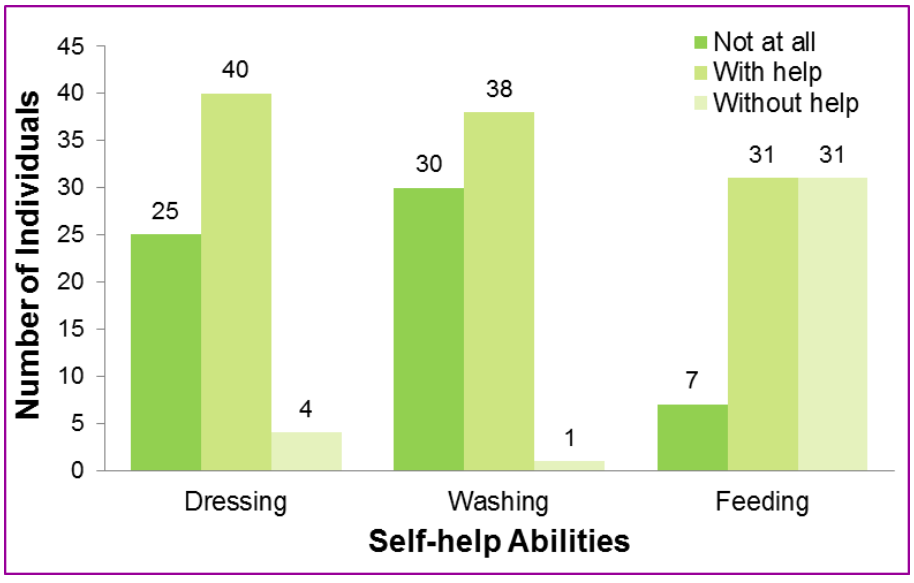

Figure 3. Reported self-help skills in individuals aged 1-36 years with SATB2-associated syndrome.

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ABILITY AND HEALTH

continued

Academic skills such as reading, writing and counting were demonstrated by some individuals with SATB2-associated syndrome (see Table 2). However, as outlined previously, 16 individuals in this sample were below five years of age. Therefore these figures should be interpreted with caution, given developmental factors and chronological age expectations.

Table 2. Academic abilities in SATB2-associated syndrome; N (%).

Reading	Writing	Counting
16 (23.19%)	17 (24.64%)	21 (30.43%)

HEALTH

Dental problems were reported by the majority of caregivers. Ear problems, cleft palate, gastrointestinal difficulties, bowel problems, epilepsy and skin problems were reported in almost half of the sample. Table 3 shows the proportion of all health-related difficulties reported in the present study.

Table 3. Health-related difficulties in SATB2-associated syndrome; N (%).

Health-related Difficulties	Number (%)
Eye problems (e.g. glaucoma/blocked tear ducts)	22 (31.9%)
Ear problems (e.g. infections, glue ear)	46 (66.7%)
Dental problems (e.g. toothache, gum problems, mouth ulcers)	66 (95.7%)
Cleft palate	33 (47.8%)
Gastrointestinal difficulties (e.g. reflux, stomach problems)	41 (59.4%)
Bowel Problems (e.g. obstruction)	31 (44.9%)
Heart abnormalities or circulatory problems (e.g. heart lesions, heart murmur)	9 (13%)
Problems with genitalia (e.g. prostate/testicular problems)	0 (0%)
Hernia (e.g. inguinal or hiatal)	2 (2.8%)
Limb abnormalities (e.g. malformed arm)	9 (13%)
Epilepsy/seizures/neurological referrals	28 (40.6%)
Lung or respiratory problems (e.g. asthma, bronchitis)	14 (20.3%)
Liver or kidney problems	5 (7.2%)
Diabetes or thyroid function problems	2 (2.8%)
Skin problems (e.g. eczema, psoriasis	30 (43.5%)

BEHAVIOUR

REPETITIVE BEHAVIOUR

The following repetitive behaviours were reported most frequently in the sample: object stereotypy (n = 32), hand stereotypy (n = 28), attachment to people (n = 34), attachment to objects (n = 34) and preference for routine (n = 27). A more detailed profile of repetitive behaviours in SATB2-associated syndrome is presented in Figure 4. A larger point towards a particular behaviour indicates that more individuals were reported as engaging in that behaviour within the previous month.

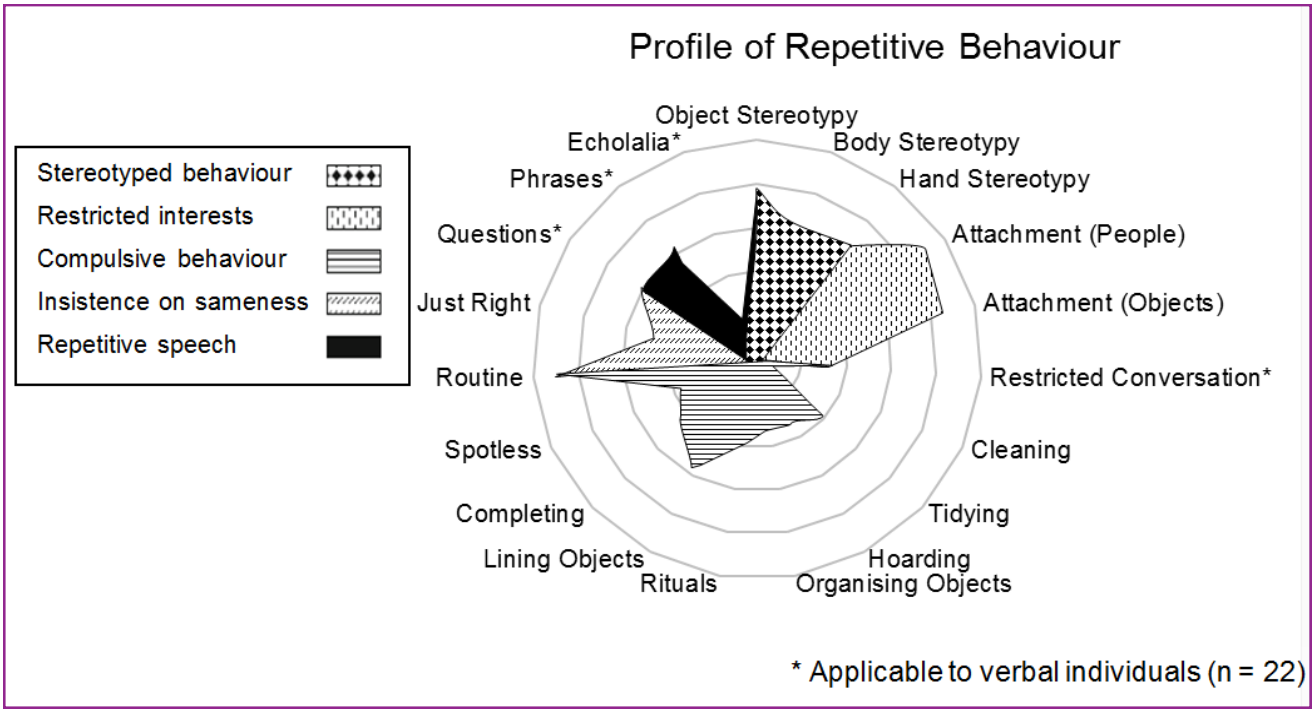

Figure 4. Profile of repetitive behaviours in SATB2-associated syndrome.

BEHAVIOUR

continued

ASD CHARACTERISTICS

Table 4 shows the number of individuals aged 4 years and older (n = 59) who scored above the clinical cut-off scores for autism spectrum disorder (ASD; score of 15 or above) and/or autism (score of 22 or above). This screening tool for ASD/autism was used in a research context and is not to be interpreted as a diagnostic measure.
For example., individuals may reach clinical cut-off scores for ASD/autism based on the number of ASD characteristics observed by their caregiver in the absence of an ASD diagnosis.

Table 4. Individuals with SATB2-associated syndrome meeting cut-off scores for ASD/autism.

Number (%) meeting cut-off for ASD	Number (%) meeting cut-off for autism
29 (49.15%)	12 (20.34%)

CHALLENGING BEHAVIOUR

The percentages of individuals with SATB2-associated syndrome presenting with self-injury, physical aggression, property destruction and stereotyped behaviour are shown in Figure 5. Based on caregiver reports, 42% of individuals evidenced self-injury, 78% evidenced physical aggression, 47% evidenced destruction of property and 34% evidenced stereotyped behaviours within the previous month.

The specific profile of self-injurious behaviours shown by the 29 individuals presenting with self-injury is outlined in more detail in Figure 6.

BEHAVIOUR

continued

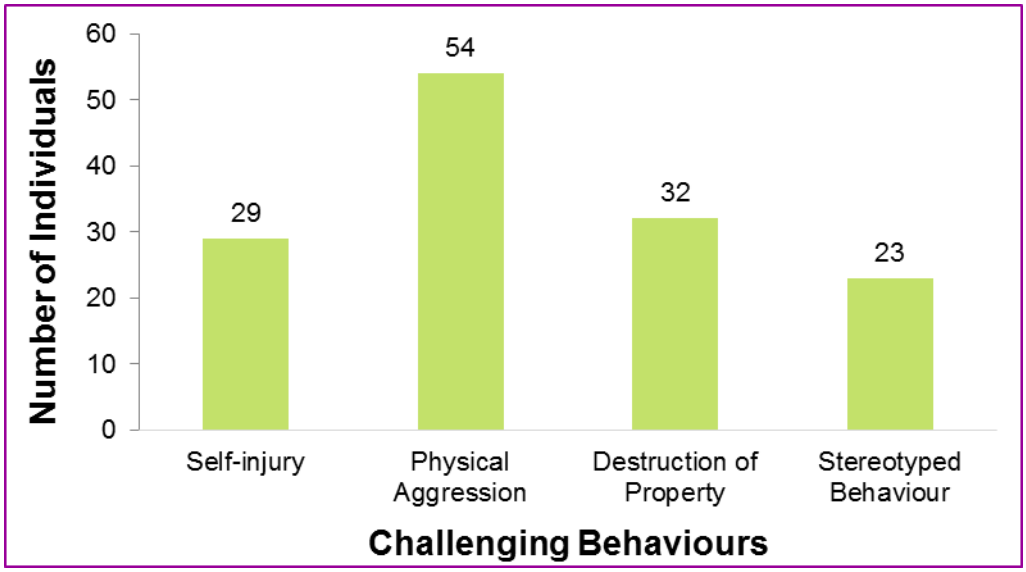

Figure 5. Profile of challenging behaviours in SATB2-associated syndrome.

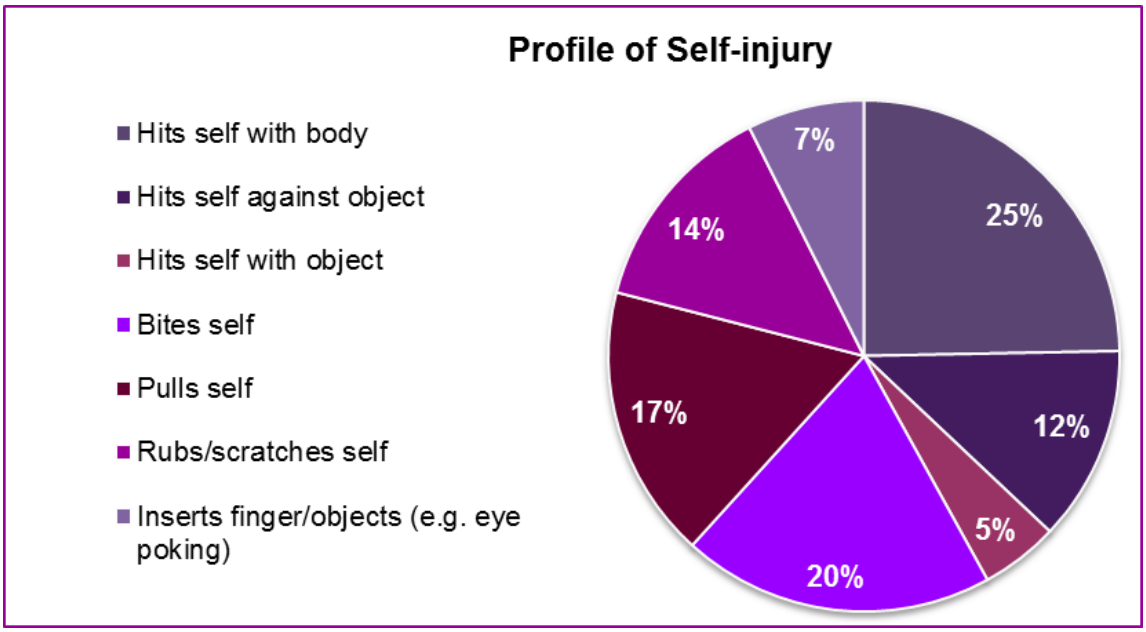

Figure 6. Profile of self-injurious behaviours in SATB2-associated syndrome (n = 29).

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This preliminary analysis provides a summary of the behavioural phenotype in SATB2-associated syndrome, based on caregiver information we have collected to date.

We sincerely hope that the content of this preliminary analysis has been useful to you. This document was prepared by Vishakha Kothari (MSc placement student, University of Birmingham) and Dr Stacey Bissell (Research Fellow, University of Birmingham). If you have any questions relating to this study or the preliminary analysis, please contact Vishakha Kothari on VXK821@student.bham.ac.uk or Dr Stacey Bissell on S.L.Bissell@bham.ac.uk.

Thank you to all the families that have taken part. Research such as this would not be possible without parents and caregivers like you. Special thanks to the SATB2 Gene Foundation for their recruitment efforts and ongoing support with our research.

For more information about behaviours in genetic syndromes, there is information available on the FIND resources website that may be useful to you. Please visit:

http://www.findresources.co.uk/common-issues

and select topics from the dropdown list available.

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