Institution: Aston University

Unit of Assessment: 3 Allied Health Professions, Dentistry, Nursing and Pharmacy

Title of case study: Improving personal and professional support for people with rare neurogenetic conditions

Period when the underpinning research was undertaken: 2017-2020

Details of staff conducting the underpinning research from the submitting unit:

Name(s):	Role(s) (e.g. job title):	Period(s) employed by submitting HEI:
Dr Jane Waite	Lecturer in Psychology	2017-date
Dr Effie Pearson	Research Associate	2020-date

Period when the claimed impact occurred: 2017-date

Is this case study continued from a case study submitted in 2014? ~~Y~~/N

1. Summary of the impact

In England alone, 370,000 people have a rare genetic syndrome that causes intellectual disability. This is a neglected population at high risk of behavioural and emotional disorder. Aston-led research has analysed profiles of behaviour in multiple rare genetic syndromes and applied the results to develop FIND (**F**urther **I**nform **N**eurogenetic **D**isorders), the first online, *accessible* resource to inform multiple stakeholders of behavioural and emotional consequences resulting from specific, rare genetic syndromes. In collaboration with support groups, FIND has significantly influenced awareness, understanding and the behaviour of parents, clinicians and educators internationally.

Impacts on **Understanding, Learning & Participation**, **Health & Wellbeing** and **Practitioners & Delivery of Professional Services** are claimed.

2. Underpinning research

Background: The White Paper 'Together for health: a strategic approach for the EU 2008-2013' and subsequent 'Recommendation on an action in the field of rare diseases' identified rare conditions as an international priority. The need for specific advice regarding behavioural profiles of genetic syndromes remains substantial. Prior research indicated that families regularly seek information tailored to their child's syndrome, but the lack of evidenced-based guidance leaves families with extreme uncertainty and places children at high risk of persistent and severe behavioural disorder. Behavioural disorder includes self-injurious behaviour (e.g. head-banging, eye gouging), aggression, rigid repetitive behaviours and life-impairing anxiety.

Behavioural disorder is a serious source of psychological distress; particularly when families are unable to understand what is happening to their loved one. A substantial proportion of parents reach cut-off for clinically significant anxiety (> 50%) and depression (> 30%). NICE guidance also emphasises how behavioural disorder impacts the person displaying that behaviour, by elevating the likelihood of children being taken into care, placement breakdown and restrictive and abusive practices. The average cost of an annual placement for someone showing behaviour disorder is £200,000.

Underpinning research: Waite's Aston-based research has elucidated the syndrome-specific behavioural profiles of eight of the ten rare genetic syndromes featured in FIND. Research into each of these syndromes was performed in collaboration with support charities (e.g. **S3.1-3.2, S3.4 & S3.6**). Updates to current knowledge regarding Cornelia de Lange (**S3.4**), Lowe, Prader-Willi, fragile-X and Williams syndromes have resulted from Aston-based research that cuts across

groups (**S3.1, S3.2-3.3, S3.5**). Further research into Rubinstein-Taybi (**S3.6**), Kleefstra, Angelman and Bardet-Biedl syndromes is ongoing in current programmes (**S3, research grants**).

Research insights/findings: Underpinning research has determined that particular genetic syndromes are at *far greater risk* than other syndromes of specific types for behaviour disorder and has identified causal mechanisms leading to these differences (**S3.1-S3.6**). This includes new insights into adaptive behaviour difficulties, severe temper-outbursts, self-injurious behaviour, aggressive behaviour, repetitive behaviours, anxiety, mood related difficulties and health/sensory difficulties across syndromes. For example, people with Williams syndrome are four times more likely to experience persistent life-limiting anxiety than people in the general population, and this is now known to be associated with underlying sensory processing problems in this syndrome (**S3.1, S3.5**).

Aston-based research also cuts across syndromes and disorders. For example, research examining mechanisms of temper outbursts in Lowe syndrome has indicated similar mechanisms as in Prader-Willi syndrome (**S3.2**), so highlighting the importance of raising awareness of findings across groups (e.g. **S3.3**), as well as within those groups.

Key Contextual Information: There is currently a lag of ~10 years between scientific publication in the field of psychology and subsequent changes to clinical practice. To address this gap, there is an urgent need for accessible summaries of research findings, training aids and resources to be made available to parents for advocacy purposes. These resources are also needed for clinicians, to accelerate the application of new knowledge in clinical settings; particularly in relation to rare and neglected groups.

3. References to the research

S3.1 Royston, R., Howlin, P., Oliver, C. & **Waite, J.** (2020). The profiles and correlates of psychopathology in adolescents and adults with Williams, fragile-X and Prader-Willi syndromes. *J. Autism Dev. Disord.* **50**, 893-903. https://doi.org/10.1007/s10803-019-04317-1

S3.2 Cressey, H., Oliver, C., & **Waite, J**. (2019). Temper outbursts in Lowe syndrome: Characteristics, sequence, environmental context and comparison to Prader-Willi syndrome. *J. Appl. Res. Intellect. Disabil.* **32**, 1216-1227. https://doi.org/10.1111/jar.12613

S3.3 Oliver, C., Dawn Adams, D., Allen, D., Crawford, H., Heald, M., Moss, J., Richards, C., Waite, J., Welham, A., Wilde, L. & Woodcock, K. (2020). The behaviour and wellbeing of children and adults with severe intellectual disability and complex needs: the BeWell checklist for carers and professionals. *Paediatrics and Child Health* **30**, 416-424. https://doi.org/10.1016/j.paed.2020.09.003.

S3.4 Groves, L., Moss., J., Crawford, H., Royston, R., **Waite, J.**, Bradley, L., Thomas, A., Moss, K., Oliver, C. (2019). The application of attentional control theory for anxiety in Cornelia de Lange syndrome. *J. Intellect. Disabil. Res.* **63**, 1081-1082. https://doi.org/10.1111/jir.12676.

S3.5 Royston, R., **Waite, J.**, & Howlin, P (2019). Williams syndrome: recent advances in our understanding of cognitive, social and psychological functioning. *Curr. Opin. Psychiatry* **32**, 60-66. https://doi.org/10.1097/YCO.0000000000000477.

S3.6 Edwards, G., Powis, L. Shelley, L., Richards, C. Oliver, C. & **Waite, J.** (2019). Repetitive behaviour in Rubinstein-Taybi syndrome: A 10-year follow-up. *J. Intellect. Disabil. Res.* **63**, 1080. https://doi.org/10.1111/jir.12676.

The quality of the above research is evidenced by **S3.1-S3.6**, published in international, peer-reviewed journals and by the following competitively-awarded research grants (PI, Waite, J.): **Baily Thomas Charitable Fund, TRUST/VC/AC/SG/4773-7697, £75,969.60**, Personal characteristics, mental health and well-being in Bardet-Biedl syndrome (2018-2022); **Lejeune Fondation, #1717, €19,993**, Ten-year longitudinal follow-up of people with Rubinstein-Taybi syndrome: Predicting mental health outcomes (2017-2019); **Autistica, Grant No. 7251, £166,000**, Towards improved

assessment of mental health difficulties in people with an Autism Spectrum Disorder, (2018-2021); **Birmingham Children's Hospital, AMacD/SP/BCHRF512, £41,000**, Anxiety in children with severe to profound intellectual disabilities, (2018-2021); **Autistica, Grant No. 7266, £98,000**, Future Leaders Award: Developing a parent-led anxiety intervention for minimally verbal autistic children, (2018-2022); and **Baily Thomas Foundation, TRUST/VC/AC/SG/5009-7975, £77,595** (2018-2021). **Cerebra** £187,133n. The Cerebra Network of Neurodevelopmental Disorders: Mental health problems in children with rare, multiple and complex needs (2020-2025).

4. Details of the impact

While a postdoc at the University of Birmingham (2014-2017, PI Prof. Chris Oliver), Waite established a new online resource, FIND (**F**urther **I**nform **N**eurogenetic **D**isorders). As an Aston academic, Waite now leads FIND which is still hosted at UoB, through an Aston/UoB collaboration. Impacts described herein are restricted to those from September 2017 onwards (i.e. Waite's Aston-based research; **S5.1**).

FIND now details ten rare genetic syndromes (**S5.2**). Four of these: Rubinstein-Taybi (RTS), Lowe, Kleefstra and Williams syndromes were developed by Waite's group at Aston, following research in collaboration with support charities (**S3.1, S3.2, S3.6**). An additional four syndromes: Angelman, Cornelia de Lange, Fragile X and Prader-Willi, have been renewed/updated by Waite since 2019, based on her cross-syndrome research (**S3.1-3.4**).

From translation of Waite's Aston research into accessible summaries, videos, assessment manuals and training aids (delivered via the FIND website) the following impacts on patients, their families and on practitioners are claimed:

Impact on understanding, learning and participation: In 2018, 87% of 104 surveyed users said they would re-visit FIND. In 2020, following Waite's updates of 2019, online feedback from 350 users revealed that 90% strongly-agreed/agreed that FIND improved their knowledge; 88% would recommend FIND to others and 91% found FIND easy to understand. Google Analytics show that FIND accumulates > 100K new web sessions per year. These are significant numbers, given the scarcity of the rare disorders addressed. FIND has global traction with key audiences comprising the United States (42%), the UK (24%), Australia (6%), Canada (6%) and India (2%) (**S5.3**). Qualitative feedback (**S5.4**) also illustrates impact e.g. *"I am a parent of a 13 year-old girl and I am also a learning disability nurse…the website has provided the most informative and accurate information I have ever read."*; "*Mother of a 9 year old. We live in the U.S. love the info here.*"; "*I'm in Japan but not enough information of PWS found in JPN website"* (parent); "*My daughter goes through bouts of aggression and I now know this is actually part of the syndrome*" and *"I'm a leader of the dutch CdLS group, can use this to help our families".* Within the UK, support groups circulate FIND via their mailing lists and estimate that between 20% and 90% of all UK families affected (according to syndrome) have been reached (**S5.5**).

Six RTS videos that Waite developed with RTS-UK detail RTS characteristics and recommendations for behaviour management (**S5.6**) and have already accumulated >2000 views via FIND (**S5.3**). Qualitative feedback demonstrates their transformative impact e.g. *"The syndrome is so rare that most of the info online is about the actual presentation and diagnosis. It was great to see the pictures/videos of others with RTS and realize that some behaviours are fairly characteristic and syndrome related. I hope to share this web-site and videos with behaviour therapists and his (special needs) school teachers"* (Canadian family, **S5.4***).*

Impact on Health and Wellbeing: Aston researchers have given invited presentations of FIND resources at 5 syndrome-support conferences, leading to parent empowerment e.g. *"They inspired me to come home and make a bigger fuss to get to the bottom of why [son] was biting himself and lashing out….– they were right, [son] had a stomach ulcer!"* and from another family: *"Information [Waite] sent to me was vital helping the authorities understand her needs and constant vulnerability for [daughter's] Personal Independent Plan assessment, and [during] a very distressing court case, the district Judge in particular took time to read it, and made good reference to it, resulting in onward support and counselling for [daughter]"* (**S5.7**). Meanwhile, qualitative feedback from the website demonstrates similar influence e.g. "*My state/country U.S. are cutting services for the disabled and*

this information is going to be so helpful in keeping the services which he presently has" (parent; **S5.4**).

Impact on Practitioners and the Delivery of Professional Services: Qualitatively, teachers have found the FIND website helpful e.g. "*sections allow us to become quickly conversant with the syndrome. The photos and videos were particularly helpful. Truly a lifesaver… for a number of us"* and "*I am from Australia and these resources will be used throughout my service"* (disability support provider) (**S5.4**). FIND is also cited in a core text and in course reading lists for trainee clinical psychologists (**S5.8**) and FIND training for these professionals has had tangible real-world benefits: e.g. "*Having this increased understanding meant the family were, as a unit, more willing to engage with me in implementing a programme to reduce [client's] repetitive behaviour"* and *"this training has made a massive difference. I feel more confident re behavioural assessments and phenotypes and will now explore the possible impacts of genetic syndromes more readily during behavioural assessments"* (**S5.9**).

In Dec-2019, a manual of 12 research questionnaires (**S5.10**), co-authored by Aston and Birmingham, was launched through FIND. The manual has already accumulated 618 downloads representing represent 37 countries, 104 NHS Trusts/organisations, 23 international hospitals/healthcare organisations, 17 government bodies and councils, 10 charities and 7 schools.

In summary, FIND addresses the 10-year lag between research and implementation by providing parents, clinicians and educators worldwide with urgently-required, up-to-date and importantly, *accessible* information and guidance about rare neurogenetic disorders.

5. Sources to corroborate the impact

S5.1 Letter from Birmingham University (collaborator) indicating Aston University's role in developing FIND and its subsequent impact.

S5.2 Further Inform Neurodevelopmental Disorders (FIND) website: www.findresources.co.uk

S5.3 Google Analytics report indicating that FIND is currently accessed internationally and that the majority of users are from outside the UK.

S5.4 Raw data/list of qualitative feedback on the FIND website collected via the ongoing, online survey and full evaluation statistics.

S5.5 Letters from five syndrome support groups estimating UK (and some international) populations reached through FIND.

S5.6 RTS videos developed at Aston University 1)https://www.findresources.co.uk/the-syndromes/rubinstein-taybi-syndrome/key-facts; 2)https://www.findresources.co.uk/the-syndromes/rubinstein-taybi-syndrome/behavioural-characteristics; 3)https://www.findresources.co.uk/the-syndromes/rubinstein-taybi-syndrome/history-prevalence; 4)https://www.findresources.co.uk/the-syndromes/rubinstein-taybi-syndrome/diagnosis; 5)https://www.findresources.co.uk/the-syndromes/rubinstein-taybi-syndrome/cognitive-characteristics;

S5.7 Email and written feedback from families and professionals who have engaged with FIND RTS resources.

S5.8 Evidence of citations of FIND in clinical training resources (scanned book insert and email evidence from course lead).

S5.9 Feedback from practising and trainee clinical psychologists who have received training using FIND resources.

S5.10 Copy of research manual that is disseminated through FIND. Oliver, C., Royston, R., Crawford, H., Moss, J., Waite, J., Arron, K., Burbidge, C., Ellis, K., Nelson, L., Ross, E., Russell, H. & Welham, A. (2019). Informant assessments of behaviour and affect for people with intellectual disability.