The British Student Doctor, 2017;1(1):17-18
doi: 10.18573/j.2017.10122
Reflections

Psychological phenotypes associated with the rare skin disease X-Linked Ichthyosis

REFLECTIONS

AUTHOR

Sohini Chatterjee
School of Medicine
Cardiff University

Address for Correspondence:
Sohini Chatterjee
Cardiff University School of Medicine, The Cochrane Building
Heath Park, Cardiff, CF14 4YU

Email: chatterjees@cardiff.ac.uk

No conflicts of interest to declare

In my intercalation year, I undertook a project looking at behavioural and psychological symptoms in a very specific group of patients affected by the rare skin condition X-linked Ichthyosis (XLI), caused by deficiency of the enzyme steroid sulfatase (STS). (1) The project was based upon previous, limited, research showing that STS plays a role in both skin and brain function, and that its deficiency can cause behavioural abnormalities. Prior to this study, evidence had only been collected from boys with XLI and animal studies. (1) I aimed to comprehensively characterise the behavioural profile for both adults and children with XLI, with the main hypothesis being that they would exhibit higher rates of symptoms related to Attention Deficit Hyperactivity Disorder (ADHD) and autism-related conditions than controls.

As XLI is a rare disease (with a prevalence rate of 1 in 3000-6000 males, (1) I undertook worldwide recruitment and testing of participants by means of an online survey. The survey link was circulated through XLI support groups and dermatological charities, taking advantage of the recent boom in using social media, such as Facebook and Twitter to obtain clinically-relevant information. We obtained ethical approval for the project through the School of Psychology, Cardiff University Ethics Committee. The study highlighted to me how even a rare, relatively inaccessible, group of patients from around the world can be recruited for a study and questioned relatively easily. (2) Overall, patients seemed keen to engage in the survey, although one of the caveats of such recruitment was response bias. Behavioural issues are not inevitable in XLI patients, but it is likely that those responding to this survey are the most affected, or over-concerned parents who may exaggerate their child's symptoms, emphasising the behavioural

issues. However, the rates of ADHD diagnosis we found were comparable with those previously seen in a sample of boys with XLI, who were ascertained based on hormonal levels and not behaviour; hence there is probably not a large response bias issue. There were also some concerns raised by affected individuals and those running support groups regarding the purpose and clinical utility of the study. Some had suspicions that this was a 'Big Pharma study' run by a pharmaceutical company for profitable intentions, which emphasised to me some of the views that the general population may harbour about online research surveys. To overcome this degree of mistrust, we explained some of the benefits of the study e.g. should the study highlight an increased prevalence of behavioural disorder traits in individuals with XLI, this may facilitate cross-speciality referrals from dermatologists and GPs (who typically diagnose XLI). Additionally, this may improve genetic counselling for the condition. With technological progress, recruiting participants internationally through online surveys will only become more prevalent, so it is our responsibility as researchers and students to make the public aware of the purpose of such research and remove any scepticism.

Our results showed that there was a substantially higher risk of neurodevelopmental disorders (particularly ADHD and autism related conditions) and associated symptoms in boys with XLI relative to unaffected controls (consistent with previous data). There is also a higher risk of both neurodevelopmental and mood disorders and associated symptoms in adults with XLI compared to unaffected controls. (3) These findings suggest that individuals with XLI are at heightened risk of psychopathology, and support the concept that the enzyme STS plays a key role in normal brain function. (3) Therefore, affected individuals may need to be referred to a multidisciplinary team including psychologists and psychiatrists, and for genetic counselling.

Importantly, our findings emphasise that there may be non-intuitive links between conditions or specialities. Many genes and proteins, including STS, act within multiple tissues of the body, and their dysfunction can elicit multiple diverse phenotypes, all of which need to be documented and treated.

I presented our results at the Ichthyosis Support Group (ISG) UK family conference, to a non-specialist patient audience consisting mainly of parents and their affected children. I acknowledged that there was a lot of complex terminology and detail involved in this project and its findings, therefore I had to take the time to explain everything clearly to help the lay audience understand the results. It was important to make parents and affected individuals aware of our results, whilst not making them overly-anxious; this was a difficult balance to achieve. We aimed to reassure participants that an XLI diagnosis was not inevitably associated with behavioural problems, but rather an increased predisposition; we emphasised that our data might allow specific impairing symptoms, such as inattentiveness and extreme hyperactivity, to be recognised early, enabling early diagnosis and treatment.

It was interesting to see many families questioning why there has not been much research on developing potential cures for ichthyotic disorders, particularly given that many are debilitating. This highlighted to me the lack of awareness and funding for rare diseases as well as the importance of support groups and charities such as the ISG in lobbying for, and providing, information and resources. The Ichthyosis Support Group itself provides research grants and is currently funding research into gene therapy for certain ichthyotic disorders. (4)

Of course, whilst I appreciate the importance of allocating limited resources to the most common conditions and those associated with the highest morbidity or mortality, just raising awareness of rare conditions amongst the GP community, specialist clinics and the general population would help greatly with diagnosis and management, and would facilitate the multidisciplinary care of those affected.

REFERENCES

1.Trent S, Davies W. Cognitive, behavioural and psychiatric phenotypes associated with steroid sulfatase deficiency. World Journal of Translational Medicine. 2013;2(1):1-12.

https://doi.org/10.5528/wjtm.v2.i1.1

2.Davies W. Insights into rare diseases from social media surveys. Orphanet Journal of Rare Disease.

https://doi.org/10.1186/s13023-016-0532-x

3. Chatterjee S, Humby T, Davies W. behavioural and psychiatric phenotypes in men and boys with x-linked ichthyosis: evidence from a worldwide online survey. PLoS One. 2016;11(10):e0164417.

https://doi.org/10.1371/journal.pone.0164417

4. Ichthyosis Support Group. Research. 2015 [accessed 18 Dec 2016]. Available from: http://www.ichthyosis.org.uk/category/research/

bsdj.org.uk


/thebsdj


@thebsdj


@thebsdj

Journal DOI
10.18573/issn.2514-3174

Issue DOI
10.18573/n.2017.10117


Cardiff University Press
Gwasg Prifysgol Caerdydd

The British Student Doctor is an imprint of Cardiff University Press, an innovative open-access publisher of academic research, where 'open-access' means free for both readers and writers.

cardiffuniversitypress.org