Young peoples’ experiences of neurofibromatosis type 1: A survey of young people, parents and health professionals

1 Centre for Appearance Research, University of the West of England, 2 Children and Family Nursing, Coventry University

Introduction

Neurofibromatosis type 1 (NF1) is a genetic condition which is highly unpredictable. The condition can result in varying degrees of visible difference (disfigurement) including café au lait spots and neurofibromas (Figures 1 and 2) which often manifest during puberty. Whilst it has been suggested that psychological problems may stem from an altered appearance caused by the condition and from the unpredictability of NF1 (Ferner et al, 2007, Ablon 1996) there is currently little research into the actual psychosocial impact of NF1 on young people. In order to aid young people in developing skills to support resilience throughout life a greater understanding of the challenges faced as well as positive adaptation of young people with NF1 is needed.

Findings

Demographics

<table>
<thead>
<tr>
<th>Young People (mean age = 20)</th>
<th>Parents</th>
<th>Health Professionals</th>
</tr>
</thead>
<tbody>
<tr>
<td>N = 73</td>
<td>N = 55</td>
<td>N = 53</td>
</tr>
<tr>
<td>66% British</td>
<td>65% British</td>
<td>57% UK based</td>
</tr>
<tr>
<td>22% N. American</td>
<td>31% N. American</td>
<td>17% N. America</td>
</tr>
<tr>
<td>12% Other</td>
<td>4% Other</td>
<td>26% Other</td>
</tr>
<tr>
<td>29% Familial NF1</td>
<td>42% had NF1 themselves</td>
<td>55% Nurses/Genetic</td>
</tr>
<tr>
<td>59% De Novo</td>
<td>44% reported</td>
<td>Counsellors</td>
</tr>
<tr>
<td>12% Unsure</td>
<td>child inherited</td>
<td>Geneticists</td>
</tr>
</tbody>
</table>

Main concerns

Managing uncertainty 44%
Having children 27%
Appearance 10%
Pain 5%
Learning difficulties 5%
Other 9%

Figure 3: Participant demographics

Health Professionals

Main concerns

Appearance 61%
Uncertainty 17%
Social 12%
Other 10%

Figure 6: Main affect of NF1 on young people

Appearance & Noticeability

There was a correlation between measures of body esteem (appearance) and measures of illness perception (B-IPQ), happiness (SHS), stigma (PSQ) and social comfort (SCQ).

<table>
<thead>
<tr>
<th>Scale</th>
<th>1 IPQ</th>
<th>2 SHS</th>
<th>3 PSQ</th>
<th>4 SCQ</th>
<th>5 BE (app.)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 IPQ</td>
<td>-0.19**</td>
<td>0.10*</td>
<td>-0.17**</td>
<td>-0.27**</td>
<td></td>
</tr>
<tr>
<td>2 SHS</td>
<td>-0.24**</td>
<td>0.26**</td>
<td>0.45**</td>
<td></td>
<td></td>
</tr>
<tr>
<td>3 PSQ</td>
<td>-0.45**</td>
<td>-0.31**</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>4 SCQ</td>
<td>-0.29**</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>5 BE (app.)</td>
<td></td>
<td></td>
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</tr>
</tbody>
</table>

Figure 5: Correlations

There was no significant difference between those who felt their NF1 was or was not noticeable to others on any of these measures except for the B-IPQ, illness perception.

Parents

Educational 25.5%
Medical 23.6%
Social 18.2%
Appearance 18.2%

Figure 6: Main affect of NF1 on child

Appearance & Noticeability

Parental reports of the noticeability of their child’s NF1 correlated with parents reports on measures related to their child’s illness threat (B-IPQ), stigma experience (PSQ) and social comfort (SCQ).

There was no correlation between parental reports of the noticeability of their child’s NF1 and parental coping.

We would like to thank all the participants for sharing their experience and also the many people and organisations who supported this study including Central Manchester University Hospitals Trust, Oxford University Hospitals NHS Trust, Great Ormond Street Hospital, The Neuro Foundation and The Children’s Tumor Foundation.

For more information please contact Jenny Barke by email: jenny.barke@uwe.ac.uk or telephone: 0117 326 1891

References

Ablon, J, Gender Response to Neurofibromatosis 1, Social Science and Medicine, 1996, 42:99–109