Simons Searchlight Registry Update HNRNPH2

April, 2022

Ages in Years

<table>
<thead>
<tr>
<th>Age Range</th>
<th>Count</th>
</tr>
</thead>
<tbody>
<tr>
<td>0-4</td>
<td>7</td>
</tr>
<tr>
<td>5-9</td>
<td>7</td>
</tr>
<tr>
<td>10-14</td>
<td>1</td>
</tr>
<tr>
<td>15-19</td>
<td>2</td>
</tr>
<tr>
<td>20-24</td>
<td>2</td>
</tr>
<tr>
<td>25+</td>
<td>1</td>
</tr>
</tbody>
</table>

Developmental and Behavioral Conditions

- Anxiety: 4
- Autism: 4
- Intellectual disability/developmental delay: 20
- Language delay: 17

Neurological Problems

- Tic disorder: 2
- Spinal cord cyst: 1
- Small head size: 10
- Seizures: 5
- Movement disorder: 6
- Low muscle tone: 18
- High muscle tone: 2
- Coordination problems: 6
- Cerebral palsy: 2

Gastrointestinal Problems

- Celiac disease: 11
- Constipation: 1
- Diarrhea: 1
- Gastric ulcer: 1
- GERD: 6

Notes:
- Graphs show counts of individuals in each category. Individual participants may appear in more than one category if they report multiple conditions.

How to Participate

The information in this report is made possible by the active participation of the HNRNPH2 community! Progress for individuals in your community with HNRNPH2 is shown below - log in to your simonssearchlight.org dashboard today to check for new surveys and tasks. Your data could hold the clues geneticists need to find answers.

STEP 1: Sign up online
STEP 2: Provide your genetic lab report
STEP 3: Share your important medical history
STEP 4: Fill out surveys
STEP 5: Provide a blood sample if you are interested
STEP 6: Update us every year

Log in to see next steps
The **Quality of Life Inventory** is a scientific survey where we asked caregivers to describe how their family member with HNRNPH2 responds to life experiences.

It measures quality of life by looking at the following areas: physical health, psychological well-being, social relationships, leisure, and independence.

These graphs reflect the number of people who reported "Often" or "Very Often" for each question.
Information Spotlight: Quality of Life

"Quality of life is a common survey of patient-centered outcomes in clinical trials and natural history studies."

- Alison Holbrook, PhD

Daily Life

- Expresses their needs: 2
- Makes own choices for activities: 3
- Likes using technology: 3
- Helps with routines: 1
- Enjoys making things: 2

Family & Friends

- Happy when they are understood: 4
- Feels relaxed while making eye contact: 3
- Initiates greetings with others: 2
- Happy to be included: 5
- Enjoys socializing at mealtimes: 3
- Enjoys interacting with others: 5
- Excited by upcoming activities: 4

Thank you for contributing knowledge to Simons Searchlight!

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