Natural History of the Refractive Errors in Patients with Congenital Stationary Night Blindness

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Introduction

• Congenital stationary night blindness (CSNB) is a group of retinal genetic disorders that primarily affect the signal transmission from the photoreceptors to the bipolar cells.1
• Complete (cCSNB) and incomplete CSNB (inCSNB) are linked to high myopia, but little is known about the natural history of refractive errors and the effect of genotype.
• This study assesses a SickKids cohort of CSNB patients to fill this knowledge gap.

Methods

• Study design: Retrospective observational longitudinal study.
• Participants: 78 patients with genetically confirmed diagnoses (CACNA1F, TRPM1, NYX, CABP4, and GPR179 mutations) of CSNB.
• Time period: from 2009 to present.
• Inclusion criteria: ≥ 2 refractive error measurements be available.
• Data collected: age, gender, CSNB sub-type (based on electroretinogram), genotype, spherical equivalent of refraction (SER), presence of nystagmus.
• Data analysis: The SER range, mean, median and rate of progression at different ages were compared between the different genotypes.

Results

<table>
<thead>
<tr>
<th>Gene</th>
<th>(Number of patients)</th>
<th>SER range across all ages.</th>
<th>Mean across all ages.</th>
<th>Median SER across all ages.</th>
<th>AGE of the youngest myopic patient / mean SER at first visit / mean SER at last visit / number of visits</th>
</tr>
</thead>
<tbody>
<tr>
<td>CACNA1F</td>
<td>(N= 52)</td>
<td>+9.00 D to 20.50 D</td>
<td>+9.45 D</td>
<td>-3.38 D</td>
<td>5 months old / first visit SER = +0.75 D OU / progressed to +9 at age of 13 years / 9 visits</td>
</tr>
<tr>
<td>TRPM1</td>
<td>(N= 14)</td>
<td>+1.50 D to +18.25 D</td>
<td>+1.90 D</td>
<td>-3.98 D</td>
<td>4 months old / first visit SER = +2.25 D OU / progressed to -8.5 OU at age of 7 years / 6 visits</td>
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<tr>
<td>NYX</td>
<td>(N= 9)</td>
<td>-1.75 D to -17.50 D</td>
<td>-10.03 D</td>
<td>-4.50 D</td>
<td>5 years old / first visit SER = -1.75 D OU / progressed to -6 at age of 10 years / 6 visits</td>
</tr>
<tr>
<td>CABP4</td>
<td>(N= 2)</td>
<td>+9.00 D to +9.50 D</td>
<td>+9.28 D</td>
<td>+3.62 D</td>
<td>30 years old / first visit SER = -7.38 D OU / progressed to -7.65 OU at age of 31 years / 2 visits</td>
</tr>
<tr>
<td>GPR179</td>
<td>(N= 1)</td>
<td>-8.25 D to -8.05 D</td>
<td>-8.09 D</td>
<td>-8.25 D</td>
<td>1 year and a half / first visit SER = -8.25 D OU / progressed to -7.80 OU at age of 2 years / 2 visits</td>
</tr>
</tbody>
</table>

Table 1: SER Range, Mean, Median across all age groups in different genotype

Conclusions

• Mean SER was different between the CACNA1F-TRPM4 related inCSNB and TRPM1-NYX related cCSNB, with the latter showing more myopia (figure 1).
• There was a statistically significant difference in the rate of SER progression per age between the two sub-groups of CSNB (P-value: 0.0143); in patients with cCSNB, the rate of SER progression of myopisation was faster (figure 2).
• Nystagmus was observed in 36/54 (66.67%) cases in cCSNB and 10/23 (43.48%) cases of cCSNB.
• A chi-square test of independence was used to examine the relationship between the CSNB groups and nystagmus. The relation between these variables was significant (P-value: 0.0285).
• Prevalence of nystagmus is unrelated to the type and degree of refractive errors.

References