

• BRIEF REPORTS •

Visual Acuity and Nystagmus following Strabismus Surgery in Patients with Oculocutaneous Albinism

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Objective: To evaluate the effect of strabismus surgery on nystagmus and visual acuity in patients with oculocutaneous albinism.

Methods: We conducted a non-concurrent retrospective study of 13 Puerto Rican patients with all types of oculocutaneous albinism who underwent strabismus surgery. Patients underwent genetic linkage analysis to reach a final oculocutaneous albinism classification prior to surgery. Strabismus surgery was modified by undercorrection of 0.5 mm in each muscle from the standard Marshall Parks' measurements in all patients.

Results: Six of the 13 patients with oculocutaneous reported an improved best corrected visual acuity after surgery. Two of the 13 patients with oculocutaneous had no nystagmus following strabismus surgery. All patients were orthotropic following surgery.

Conclusion: Strabismus surgical undercorrection may be of benefit in patients with oculocutaneous albinism. [*P R Health Sci J* 2010;4:391-393]

Key words: Nystagmus, Oculocutaneous albinism, Strabismus

Oculocutaneous albinism (OCA) is a group of genetic disorders with a wide phenotypic variability in which patients have a decreased melanin pigmentation in the eyes, skin, and hair (1). Parks and co-workers suggest that a defective tyrosinase enzyme system leads to developmental abnormalities in the neural tissue (1-2). Recent studies have elucidated via genetic linkage analysis the genes responsible for different types of OCA (3).

The most common ophthalmic findings in patients with OCA include decreased visual acuity (VA), nystagmus, strabismus, iris transillumination, foveal hypoplasia, albinotic retinal mid-periphery and optic track misrouting (1-6).

Previous studies suggest that bilateral extraocular muscle recession may improve visual acuity and decrease nystagmus (7-9). Davis and co-workers have suggested that four-muscle horizontal rectus recessions may improve unocular and binocular visual acuities in albinos with nystagmus as measured by Snellen chart (10). Previous clinical experience by the one of the authors suggested that the standard surgical correction of strabismus as described by Marshall Parks in patients with albinism resulted in a significant increase in surgical overcorrection of strabismus.

Several types of OCA have been described in Puerto Rico (PR). However, in PR the most common type of OCA is the Hermansky-Pudlak syndrome (HPS) (11). Patients with the HPS have a triad of tyrosinase-positive oculocutaneous

albinism, bleeding diathesis and accumulation of ceroid in tissues (1-6,12-15).

The purpose of this study was to investigate if strabismus surgery improves VA and nystagmus in Puerto Rican patients with OCA. Furthermore, this study evaluated the surgical outcome of undercorrection of heterotropia in patients with OCA.

Patients and Methods

We conducted a non-concurrent retrospective study of 13 Puerto Rican patients with all types of OCA who underwent strabismus surgery. All patients underwent a comprehensive systemic and ophthalmic evaluation prior to and one day, one week, and one month following strabismus surgery in the San Juan metropolitan area, from 1998 to 2006. Patients were also evaluated yearly after the surgical correction was performed.

Best corrected visual acuity (BCVA) was evaluated monocularly using a +5.00 sphere lens in the contralateral eye, using a Snellen chart at 20 feet and at two feet distance for those patients with low vision. Nystagmus was evaluated and classified in terms of

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type, frequency and amplitude. Strabismus was evaluated with correction and using a modified Krinsky technique.

Patients underwent genetic linkage analysis at John Hopkins Hospital to reach a final OCA classification prior to surgery. Standard Marshall Parks' strabismus surgery was performed with a modified under-correction of 0.5 mm in each muscle from the standard Marshall Parks' measurements in all patients (16-17). Post-operative follow-up visits included visual acuity, nystagmus, and strabismus evaluation. Student t-test analyses were performed to compare pre-operative and post-operative BCVA, nystagmus, and heterotropia.

Results

There were four male and nine female patients. Age ranged from 3 to 25 years (eight patients < 8 years, five patients > 5 years) with a mean [standard deviation (SD)] age of 6.0 (6.0) years. The follow up time ranged from 0.2 to 6 years with a mean (SD) follow up time of 1.4 (1.7) years. Eight patients had oculocutaneous albinism and five had HPS.

Pre-operative BCVA of the right eye ranged from 20/30 to 20/400 with a mean (SD) of 20/225.4 (20/155.0). Pre-operative BCVA of the left eye ranged from 20/30 to 20/400 with a mean (SD) of 20/240.8 (20/162.6). The pre-operative VA of all eyes ranged from 20/30 to 20/400 with a mean of 20/233.1 (20/155.6). Post-operative BCVA of the right eye ranged from 20/40 to 20/400 with a mean of 20/198.5 (20/133.7). Post-operative BCVA of the left eye ranged from 20/30 to 20/400 with a mean (SD) of 20/197.7 (20/134.5). Post-operative BCVA of all eyes ranged from 20/30 to 20/400 with a mean (SD) of 20/198.01 (20/131.5).

Six of the 13 patients (46%) with OCA reported an improved BCVA after surgery. Three out of 13 patients (23%) reported a diminished BCVA. Four out of 13 patients (31%) reported no change in VA after surgery.

Of those patients with HPS, two out of five patients (40.0%) reported an improved BCVA. One out of five patients (20.0%) with the HPS reported no change in BCVA following surgery. Two patients out of five (40%) with the HPS reported a decrease in BCVA following surgery.

All patients had congenital nystagmus. Three patients (23.1%) had rotatory nystagmus. Ten patients (76.9%) had periodic alternating nystagmus. Nystagmus' frequency varied from slow to fast, and amplitude ranged from small to wide. Two of the 13 (15.4%) patients with OCA had no nystagmus following strabismus surgery. Nystagmus following surgery was found in 11 out of 13 patients (84.6%).

Five out of 13 patients (38.5%) with OCA had congenital exotropia. Eight out of 13 patients (61.5%) had congenital esotropia. Pre-operative heterotropia of all eyes ranged from 15 to 45 prism diopters with a mean (SD) of 30.9 (9.4). All patients with esotropia underwent bilateral medial rectus recessions.

Three patients underwent bilateral lateral rectus recessions. All patients were orthotropic following surgery.

Statistical analyses showed a significant difference ($p = 0.003$) between pre-operative and post-operative heterotropia as measured by prism diopters. No significant changes were found for BCVA and nystagmus.

Discussion

Strabismus surgery in patients with OCA remains a challenge. Previous studies have reported that four-muscle strabismus surgery may improve BCVA in patients with OCA (10). Bagheri and coworkers have reported improvement in BCVA following strabismus surgery in up to 76.5% of patients (7). In our study, 46% of patients with OCA had an improved BCVA following strabismus surgery. These findings are similar to those reported by Davis and co-workers (10). However, a difference between pre-operative and post-operative VA in our study was not found to be statistically significant. The difference in BCVA prognosis between patients with OCA and the general population following strabismus surgery appears to be due to foveal hypoplasia in patients with OCA (15). Further studies that include pre-operative macular optical coherence tomography may elucidate a relationship between BCVA prognosis in patients with OCA after strabismus surgery to the degree of foveal hypoplasia.

Previous studies suggest that patients with oculocutaneous albinism without nystagmus can be expected to demonstrate better VA compared with those with obvious nystagmus (18-19). In our study, 92.3% of patients had nystagmus. This may explain a poorer BCVA prognosis in our patients.

Davis and colleges suggest that rectus muscle surgery may improve nystagmus in patients with oculocutaneous albinism (10). Our study did not find a statistically significant difference between pre-operative and post-operative nystagmus. Previous studies report patient qualitative improvement in nystagmus after strabismus surgery even if nystagmus is present post-operatively in the physical examination (7). Further studies need to be undertaken to investigate if Puerto Rican patients with oculocutaneous albinism that have nystagmus report clinical improvement after strabismus surgery regardless of physical findings at examination.

This study found a statistically significant difference between pre-operative and post-operative heterotropia as measured by prism diopters. The surgical procedure used in our study undercorrects the heterotropia by 0.5 mm when compared to the Marshall Parks' technique. Although all patients were undercorrected surgically, none had heterotropia post-operatively. This surgical phenomenon may be related to the abnormal neurologic development seen in patients with all types of albinism. Further studies may elucidate a table of reference to surgically correct patients with oculocutaneous albinism.

To our knowledge, this study is the first report to date that evaluates if strabismus surgery improves VA, nystagmus, and heterotropia in Puerto Rican patients with OCA and patients with the HPS. Studying the post-operative results of strabismus surgery on patients with OCA may lead to better management strategies for this population. This is especially important in patients with bleeding diathesis such as patients with the HPS.

Limitations in our study include a small sample size, lack of long-term follow up, and high prevalence of HPS in our study population. Further studies need to be done that screen pre-operatively with OCT for macular anomalies to eliminate the confounding factor of macular hypoplasia in VA prognosis.

In conclusion, our study highlights some characteristics of albinos that have undergone strabismus surgery in Puerto Rico. Our study population did not show a statistically significant improvement in VA or nystagmus. However, our study accentuates the need to delineate that different types of oculocutaneous albinism might benefit from different management strategies. Strabismus surgical undercorrection may be of benefit in patients with OCA. Further studies with standardized controls and a larger sample size are needed to evaluate the population of albinos with strabismus living in Puerto Rico.

Resumen

Objetivo: El objetivo del estudio es evaluar el efecto que tiene la cirugía de estrabismo en pacientes con albinismo oculocutáneo en términos de la agudeza visual y nistagmo. **Métodos:** Este es un estudio retrospectivo de trece pacientes puertorriqueños con albinismo oculocutáneo de todos los tipos que se sometieron a cirugía de estrabismo. Se clasificó genéticamente los pacientes con albinismo oculocutáneo previo a cirugía. La cirugía de estrabismo fue modificada en todos los pacientes para corregir cada músculo 0.5 mm menos de las medidas estándar de Marshall Parks. **Resultados:** Seis de los 13 pacientes con albinismo oculocutáneo mejoraron su visión corregida después de la cirugía de estrabismo. Dos de los 13 pacientes con albinismo oculocutáneo tuvieron ausencia de nistagmo después de la cirugía de estrabismo. Todos los pacientes

estuvieron ortotrópicos después de la cirugía. **Conclusión:** Corregir cada músculo 0.5 mm menos de las medidas estándar de Marshall Parks durante la cirugía de estrabismo puede ser beneficioso en pacientes con albinismo oculocutáneo.

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