


Neurodevelopmental Profile in Children Affected by Ocular Albinism

Jessica Galli^{1,2}  Erika Loi^{2,3} Alessandra Morandi^{1,2} Vera Scaglioni² Andrea Rossi² Anna Molinaro² Nadia Pasini⁴ Francesco Semeraro⁵ Giulio Ruberto⁶ Elisa Fazzi^{1,2}

¹Department of Clinical and Experimental Sciences, University of Brescia, Brescia, Italy

²Unit of Child Neurology and Psychiatry, ASST Spedali Civili of Brescia, Brescia, Italy

³Department of Molecular and Translational Medicine, University of Brescia, Brescia, Italy

⁴Department of Neurological and Vision Sciences, ASST Spedali Civili of Brescia, Eye Clinic, Brescia, Italy

Address for correspondence Jessica Galli, MD, PhD, Unit of Child Neurology and Psychiatry, ASST Spedali Civili of Brescia, Piazzale Spedali Civili, 1, 25123, Brescia, Italy (e-mail: jessica.galli@unibs.it).

⁵Department of Neurological and Vision Sciences, ASST Spedali Civili of Brescia and University of Brescia, Eye Clinic, Brescia, Italy

⁶Department of Surgical and Clinical, Diagnostic and Pediatric Sciences, Section of Ophthalmology, University of Pavia, IRCCS Fondazione Policlinico San Matteo, Pavia, Italy

Neuropediatrics

Abstract

Aim The aim of this study was to detail the neurodevelopmental profile of subjects affected by ocular albinism (OA) and to collect data on GPR143 gene analysis.

Design The design of the study involves a retrospective longitudinal observational case series.

Methods We collected data on the neurodevelopmental profile of 13 children affected by OA from clinical annual assessments conducted for a period of 6 years after the first evaluation. We described visual profile, neuromotor development and neurological examination, cognitive profile, communication and language skills and behavioral characteristics. The GPR143 gene analysis was performed as well.

Results Children presented a variable combination of ocular and oculomotor disorders unchanged during the follow-up, a deficit in visual acuity and in contrast sensitivity that progressively improved. Abnormalities in pattern visual evoked potential were found. No deficits were detected at neurological examination and neuromotor development except for a mild impairment in hand-eye coordination observed in five cases. A language delay was observed in five cases, two of whom had also a developmental quotient delay at 2 years evolving to a borderline/deficit cognitive level at preschool age, difficulties in adaptive behavior and autistic-like features were found. Mutations in the GPR143 gene were identified in the two patients who presented the most severe clinical phenotype.

Conclusion Children with OA may share, in addition to a variable combination of ocular signs and symptoms, a neurodevelopment impairment regarding mostly the cognitive, communicative, and social area, especially those with GPR143 mutation.

Acknowledgments

The authors are grateful to Anna Alessandrini, Alice Bertoletti, Nicole D'Adda, Alessandra Franzoni, and Melissa Marras for their valuable help in evaluating the children included in this study. We thank children affected by ocular albinism and their care providers for participating and Albinet APS as well.

Keywords

- ▶ ocular albinism
- ▶ neurodevelopmental profile
- ▶ autism spectrum disorder
- ▶ GPR143 gene mutations