

# **Multimodal phenotyping of foveal hypoplasia in albinism and albino-like conditions: a pediatric case series with adaptive optics insights**

Bacci Giacomo Maria<sup>1\*</sup> MD PhD, Marziali Elisa<sup>1\*</sup> MD, Bargiacchi Sara<sup>2</sup> MD, Paques Michel<sup>3,4</sup> MD PhD, Virgili Gianni<sup>5,6</sup> MD, Fortunato Pina<sup>1</sup> MD PhD, Durand Marine<sup>7</sup>, Rocca Camilla<sup>8</sup> MD, Pagliuzzi Angelica<sup>9</sup> MD, Palazzo Viviana<sup>2</sup>, Tiberi Lucia<sup>2</sup>, Vergani Debora<sup>2</sup>, Landini Samuela<sup>2</sup>, Peron Angela<sup>2,8</sup> MD PhD, Artuso Rosangela<sup>2</sup> PhD, Pacini Bianca<sup>5</sup> MD, Stabile Monica<sup>10</sup> MD, Sodi Andrea<sup>5</sup> MD, Caputo Roberto<sup>1</sup> MD

1: Pediatric Ophthalmology Unit A. Meyer Children's Hospital IRCCS, Florence, Italy

2: Medical Genetics Unit, Meyer University Hospital IRCCS, Florence, Italy

3: Clinical Investigation Center Vision 1423, INSERM-DGOS, Sorbonne Université, Quinze-Vingts Hospital, Paris, France.

4: Institut de la Vision, Paris, France.

5: Department of Neuroscience, Psychology, Drug Research and Child Health, University of Florence, Florence, Italy.

6: IRCCS – Fondazione Bietti, Rome, Italy.

7: Imagine Eyes, Orsay, France

8: Department of Biomedical Experimental and Clinical Sciences "Mario Serio", University of Florence, Florence, Italy.

9: Nephrology and Renal Transplantation Research Group, KU Leuven, Belgium

10: Department of Clinical and Experimental Medicine, Section of Pediatrics, University of Pisa, Pisa, Italy

\*These authors equally contributed

<b>Gene panel</b>	
<i>AHR</i>	NM_001621.5
<i>AP3B1</i>	NM_003664.5
<i>AP3D1</i>	NM_001261826.3
<i>BLM</i>	NM_000057.4
<i>BLOC1S3</i>	NM_212550.5
<i>BLOC1S5</i>	NM_201280.3
<i>BLOC1S6</i>	NM_012388.4
<i>CACNA1A</i>	NM_001127221.2
<i>CACNA1F</i>	NM_005183.4
<i>CASK</i>	NM_003688.3
<i>DTNBP1</i>	NM_032122.5
<i>EDN3</i>	NM_207034.3
<i>EDNRB</i>	NM_000115.5
<i>FRMD7</i>	NM_194277.3
<i>GNAI3</i>	NM_006496.4
<i>GPR143</i>	NM_000273.3
<i>HPS1</i>	NM_000195.5
<i>HPS3</i>	NM_032383.5
<i>HPS4</i>	NM_022081.6
<i>HPS5</i>	NM_181507.2
<i>HPS6</i>	NM_024747.6
<i>KITLG</i>	NM_000899.5
<i>KIT</i>	NM_000222.3
<i>LRMDA</i>	NM_032024.5
<i>LYST</i>	NM_000081.4
<i>MANBA</i>	NM_005908.4
<i>MITF</i>	NM_000248.4
<i>MLPH</i>	NM_024101.7
<i>MYO5A</i>	NM_000259.3
<i>OCA2</i>	NM_000275.3
<i>PAX3</i>	NM_181457.4
<i>PAX6</i>	NM_000280.6
<i>RAB27A</i>	NM_004580.5
<i>SACS</i>	NM_014363.6
<i>SETX</i>	NM_015046.7
<i>SLC24A5</i>	NM_205850.3
<i>SLC38A8</i>	NM_001080442.3
<i>SLC45A2</i>	NM_016180.5
<i>SNAI2</i>	NM_003068.5
<i>SOX10</i>	NM_006941.4
<i>TULP1</i>	NM_003322.6
<i>TYRP1</i>	NM_000550.3
<i>TYR</i>	NM_000372.5

**Supplementary Table S1.** Panel of genes associated with foveal hypoplasia and analysed by Exome Sequencing