Supplemental Table S1. Patient demographics and genetic testing results.

| Patient # | Race and Ethnicity | Cutanous Findings | Ocular Findings | Family History | Genetic Test | Date of Test | Gene (Variant Classification) | Diagnostic Yield Positive |
|---|---|---|---|---|---|---|---|---|
| **1** | Other/ Hispanic | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia | No | Albinism Panel (Dermatology) Plus | 2021 | *OCA2* (P) | No |
| **2** | White/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia | Yes (related to patient 3) | Albinism Panel (Dermatology) Plus | 2019 | *HPS5* (LP) | Yes |
| **3** | Other/NH | Yes | Nystagmus, foveal hypoplasia | Yes (related to patient 2) | Familial Variant Testing | 2019 | *HPS5* (LP) | Yes |
| **4** | Other/ Hispanic | Yes | Nystagmus, foveal hypoplasia, iris TID | No | DNA Analysis for Mutations in the Human HPS-1 and HPS-3 genes | 2015 | *HPS1* (LP/P) | Yes |
| **5** | White/NH | No | Nystagmus, foveal hypoplasia | No | Albinism Panel (Dermatology) Plus | 2018 | *TYR* (P), *TYR* (P), *LYST* (VUS) | Yes |
| **6** | Unknown | No | Nystagmus, fundus hypopigmentation, foveal hypoplasia, iris TID | No | Hypopigmentation Panel | 2021 | *HPS6* (LP) | Yes |
| **7** | Other/ Hispanic | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia, iris TID | No | Oculocutaneous Albinism Panel | 2019 | *TYR* (P), *TYR* (P) | Yes |
| **8** | Black/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia, iris TID | Yes | Oculocutaneous Albinism Panel | 2021 | *SLC45A2* (P), *SLC45A2* (P) | Yes |
| **9** | Declined | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia, iris TID | No | Whole Exome Sequencing | 2020 | *TYR* (P), *TYR* (P) | Yes |
| **10** | Other/ Hispanic | No | Nystagmus, foveal hypoplasia | Yes | GPR143 DNA Analysis | 2009 | No mutation | No |
| **11** | Other/ Hispanic | Yes | Nysatgmus, fundus hypopigmentation, foveal hypoplasia, iris TID | Yes | Albinism Panel (Dermatology) Plus | 2021 | *OCA2* (P), *OCA2* (LP) | Yes |
| **12** | Black/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia, iris TID | No | Albinism Panel (Dermatology) Plus | 2020 | *OCA2* (P), *OCA2* (LP) | Yes |
| **13** | Black/NH | Yes | None | No | Albinism Panel by Massively Parallel Sequencing | 2015 | *OCA2* (P) | No |
| **14** | White/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia, iris TID | No | Albinism Panel by Massively Parallel Sequencing | 2020 | *TYR* (P), *TYR* (P) | Yes |
| **15** | Black/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia, iris TID | Yes | OCA2 Targeted Deletion Analysis | 2012 | *OCA2* (LP/P), *OCA2* (LP/P) | Yes |
| **16** | Other/ Hispanic | No | Nystagmus, foveal hypoplasia, iris TID | No | Common Mutations in HPS1,HPS3/Hermansky-Pudlak Syndrome, TYR, OCA2 | 2006 | *OA1/GPR143* (VUS->P (reclassification)), *TYR* (VUS/risk factor ->VUS/risk factor (reclassification)) | No->Yes |
| **17** | Other/His panic | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia | Yes | Hypopigmentation Panel | 2021 | *OCA2* (P), *OCA2* (VUS), *AP3D1* (VUS), *TYR* (VUS), *HPS5* (VUS) | Yes |
| **18** | Other/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia | No | Exomenext Select | 2019 | *OCA2* (P), *OCA2* (LP), *MC1R* (VUS) | Yes |

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| **19** | White/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia | No | Invitae Inheirted Retinal Disorders Panel | 2021 | *OCA2* (P), *CNGA3* (VUS), *CYP4V2* (VUS), *GPR179* (VUS), *RP1* (VUS) | No |
| **20** | White/NH | Yes | Nystagmus, foveal hypoplasia | Yes | Oculocutaneous Albinism Panel | 2020 | *OCA2* (P), *OCA2* (LP), *TYR* (LP) | Yes |
| **21** | Native Hawaiian or Pacific Islander/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia, iris TID | No | Albinism Panel by Massively Parallel Sequencing | 2014 | *OCA2* (LP/P), *OCA2* (LP/P), *HPS5* (LP/P) | Yes |
| **22** | White/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia | No | Albinism Panel (Ophthalmology) Plus | 2018 | *TYR* (P), *TYR* (VUS) | Yes |
| **23** | Other/Hispanic | Yes | Nystagmus, foveal hypoplasia | No | NGS Pigmentation SmartPanel Casey Eye | 2015 | *TYRP1* (LP/P), *TYRP1* (LP/P) | Yes |
| **24** | White/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia, iris TID | Yes | Hermansky Pudlack and Oculocutaneous Albinism Panel | 2015 | *OCA2* (LP/P), *OCA2* (LP/P), *TYRP1* (LP/P), *LYST1* (nonsense mutation) | Yes |
| **25** | White/NH | Yes | Nystagmus, foveal hypoplasia | Yes | Albinism Panel (Dermatology) Plus | 2018 | *OCA2* (P), *OCA2* (LP) | Yes |
| **26** | White/NH | Yes | Nystagmus, fundus hypopigmentation | No | Albinism Panel (Dermatology) Plus | 2018 | *TYR* (VUS->VUS (reclassification)) | No |
| **27** | Black/NH | Yes | Nystagmus | Unknown | Hermansky Pudlok Syndrome, Type I, Sequence | 2008 | No mutation | No |
| **28** | White/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia, iris TID | No | Oculocutaneous Albinism Panel | 2021 | *TYR* (P), *TYR* (VUS->P (reclassification)), *TYR* (VUS), *RAB27A* (VUS), *MC1R* (VUS) | No->Yes |
| **29** | Asian/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia | Unknown | Oculocutaneous Albinism Panel, 5 Genes, Casey Eye Institute | 2012 | *OCA2* (VUS->VUS (reclassification)), *OCA2* (VUS->VUS (reclassification)) | No |
| **30** | White/NH | Yes | Fundus hypopigmentation, iris TID | No | Oculocutaneous Albinism Panel | 2012 | *TYR* (LP/P), *TYR* (LP/P) | Yes |
| **31** | White/NH | No | Nystagmus, fundus hypopigmentation, foveal hypoplasia | Yes | Albinism Sequence Analysis | 2016 | *OA1/GPR143* (VUS->VUS (reclassification)), *OCA2* (VUS->VUS (reclassification)) | No |
| **32** | White/NH | Yes | Fundus hypopigmentation, foveal hypoplasia, iris TID | Unknown | Hermansky Pudlak and Oculocutaneous Albinism Panel | 2013 | *OCA2* (LP/P), *OCA2* (LP/P) | Yes |
| **33** | White/Hispanic | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia | Yes | Oculocutaneous Albinism Panel | 2020 | *OCA2* (P), *OCA2* (P), *GPR143* (VUS) | Yes |
| **34** | White/NH | No | Nystagmus, fundus hypopigmentation, foveal hypoplasia | Yes | Oculocutaneous Albinism Sequencing Panel | 2017 | *OCA2* (LP), *TYR* (VUS->haplotype | No |

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| | | | | | | | (reclassification)), *TYR* (VUS) | |
| **35** | White/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia | No | ID my IRD panel | 2021 | *TYR* (P), *TYR* (LP) | Yes |
| **36** | White/NH | Yes | Nystagmus, foveal hypoplasia | No | Oculocutaneous Albinism Panel | 2021 | *TYR* (P), *TYR* (P) | Yes |
| **37** | Black/NH | Yes | Nystagmus, foveal hypoplasia | Yes | Exome Next Select for OCA Genes | 2019 | *HPS5* (P), *OCA2* (LP), *OCA2* (VUS) | Yes |
| **38** | White/NH | No | Foveal hypoplasia, iris TID | No | Invitae Inherited Retinal Disorders Panel | 2021 | *TYR* (P), *MPDZ* (P), *CDH23* (VUS), *CTNNA1* (VUS), *HMX1* (VUS), *LRP2* (VUS), *ZNF423* (VUS) | No |
| **39** | Unknown | No | Nystagmus, fundus hypopigmentation, foveal hypoplasia | No | Invitae Inherited Retinal Disorders Panel + WES/Mitochondrial | 2021 | *OCA2* (P), *ARHGEF18* (VUS), *IDH3A* (VUS), *KIF7* (VUS), *MTTP* (VUS), *SLC7A14* (VUS) | No |
| **40** | White/NH | No | Nystagmus, foveal hypoplasia | No | Oculocutaneous Albinism Sequencing Panel | 2017 | *TYR* (VUS), *TYR* (VUS) | No |
| **41** | Other/Hispanic | Yes | Fundus hypopigmentation, iris TID | Unknown | Albinism Panel by Massively Parallel Sequencing | 2014 | *OCA2* (LP/P), *HPS6* (VUS->VUS (reclassification)) | No |
| **42** | Unknown | Yes | Nystagmus | Yes | Oculocutaneous Albinism Panel | 2020 | *TYRP1* (P) | Yes |
| **43** | Other/Hispanic | Yes | Nystagmus, foveal hypoplasia | No | Albinism Panel (Dermatology) Plus | 2018 | *TYR* (P), *TYR* (VUS), *HPS4* (VUS), *HPS6* (VUS) | No |
| **44** | White/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia, iris TID | No | Oculocutaneous Albinism Panel | 2019 | *TYR* (P), *TYR* (VUS), *TYR* (VUS) | Yes |
| **45** | Black/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia | Unknown | Hermansky Pudlak and OCA 15 Gene Panel | 2016 | *OCA2* (LP/P), *OCA2* (LP/P) | Yes |
| **46** | Other/Hispanic | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia, iris TID | No | HPS1 and HPS3 Genes/ Select Analysis of Common Mutations / Hermansky-Pudlak Syndrome | 2008 | *HPS5* (LP/P), *HPS5* (LP/P) | Yes |
| **47** | White/NH | No | Nystagmus, fundus hypopigmentation, foveal hypoplasia | No | Pigmentation Panel through Casey Eye Institute | 2014 | *TYR* (LP/P), *TYR* (LP/P), *TYR* (VUS), *HPS5* (LP/P) | Yes |
| **48** | Other/Hispanic | Yes | Nystagmus, foveal hypoplasia, iris TID | Yes | Oculocutaneous Albinism Panel | 2019 | *TYR* (P), *OCA2* (VUS) | Yes |
| **49** | Black/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia | No | ExomeNext-Select: Analysis of Medical Provider Selected Genes | 2020 | *OCA2* (P), *OCA2* (LP) | Yes |
| **50** | Other/Hispanic | No | Nystagmus, fundus hypopigmentation | No | Ocular Albinism, X-linked GPR143 Comprehensive - Sequence & Deletion/Duplication Analysis | 2015 | *OA1/GPR143* (P) | Yes |
| **51** | Black/NH | Yes | Fundus hypopigmentation | No | Hermansky Pudlak Syndrome and Oculocutaneous Albinism, CEI | 2015 | *OCA2* (LP/P), *OCA2* (LP/P) | Yes |

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|---|---|---|---|---|---|---|---|---|
| | | | | | Molecular Diagnostics, RT / NGS Pigmentation SmartPanel | | | |
| **52** | White/NH | No | Nystagmus, fundus hypopigmentation, foveal hypoplasia | No | Blueprint Genetics FLEX Albinism Panel (Ophthalmology) Plus | 2022 | No mutation | No |
| **53** | White/NH | Yes | Nystagmus, fundus hypopigmentation, foveal hypoplasia | Yes | Invitae Inherited Retinal Disorders Panel | 2022 | *CACNA2D4* (VUS), *COL18A1* (VUS), *FSCN2* (VUS), *PCDH15* (VUS), *RIMS1* (VUS) | No |

"NH" = Not Hispanic. "N/A" = Not Available. "TID" = Transillumination Defect. "P" = Pathogenic. "LP" = Likely Pathogenic. "VUS" = Variants of Uncertain Significance.
"Yes" for Cutaneous findings if characteristic skin findings of albinism were seen
"TID" = transillumination defects
"Yes" for Family History includes patients with family history of cutaneous or ocular manifestations of albinism.
"Yes" for positive diagnostic yield indicates that the patient had genetic testing results that are believed to explain the patient's ocular or oculocutaneous albinism based on the variant classification and inheritance pattern.
Patient 16 and 28 had reclassifications of VUS' to P variants that changed the diagnostic yield, Other VUS that underwent reclassification that remained VUS are also listed with "VUS->VUS (reclassification)"