Mild form of oculocutaneous albinism type 1: phenotypic analysis of compound heterozygous patients with the R402Q variant of the gene

Titre	Mild form of oculocutaneous albinism type 1: phenotypic analysis of compound heterozygous patients with the R402Q variant of the gene
Type de publication	Article de revue
Auteur	Monfermé, Solene [1], Lasseaux, Eulalie [2], Duncombe-Poulet, Catherine [3], Hamel, Christian [4], Defoort-Dhellemmes, Sabine [5], Drumare, Isabelle [6], Zanlonghi, Xavier [7], Dollfus, Hélène [8], Perdomo, Yaurama [9], Bonneau, Dominique [10], Korobelnik, Jean-François [11], Plaisant, Claudio [12], Michaud, Vincent [13], Pennamen, Perrine [14], Rooryck-Thambo, Caroline [15], Morice-Picard, Fanny [16], Paya, Clement [17], Arveiler, Benoit [18]
Editeur	BMJ Publishing Group
Type	Article scientifique dans une revue à comité de lecture
Année	2018
Langue	Anglais
Date	24 Nov. 2018
Titre de la revue	British journal of ophthalmology
ISSN	1468-2079
Résumé en anglais	AIM: Oculocutaneous albinism type 1 (OCA1) is due to mutations. c.1205G>A/p.Arg402Gln (R402Q) is a thermosensitive variant of the gene that has been reported to be responsible for mild forms of OCA1. The aim of our study was to define the phenotype associated with this variant. METHODS: In our retrospective series, among 268 patients diagnosed with OCA1, 122 (45.5%) harboured one pathogenic variant of , and the R402Q variant ensured to be in trans by segregation analysis in 69 patients (25.7%), constituting the 'R402Q-OCA1' group. 146 patients harboured two pathogenic variants of the gene other than R402Q. Clinical records were available for 119 of them, constituting the 'Classical-OCA1' group. RESULTS: Most R402Q-OCA1 patients presented with white or yellow-white hair at birth (71.43%), blond hair later (46.97%), a light phototype but with residual pigmentation (69.64%), and blue eyes (76.56%). Their pigmentation was significantly higher than in the classical-OCA1 group. All patients from the R402Q-OCA1 group presented with ocular features of albinism. However the prevalence of photophobia (78.13%) and iris transillumination (83.87%) and the severity scores of iris transillumination, retinal hypopigmentation and foveal hypoplasia were lower in the R402Q-OCA1 group. Visual acuity was higher in the R402Q-OCA1 group (0.38±0.21 logarithm of the minimum angle of resolution vs 0.76±0.24). Investigations concerning a possible additive effect of the c.575C>A/p.Ser192 (S192Y) variant of in cis with R402Q, suggested by others, showed no significant impact on the phenotype. CONCLUSION: The R402Q variant leads to variable but generally mild forms of albinism whose less typical presentation may lead to underdiagnosis.

URL de la notice	http://okina.univ-angers.fr/publications/ua18418 [19]
DOI	10.1136/bjophthalmol-2018-312729 [20]
Lien vers le document	https://bjo.bmj.com/content/early/2018/11/24/bjophthalmol-2018-312729 [21]
Titre abrégé	Br J Ophthalmol
Identifiant (ID) PubMed	30472657 [22]

Liens
[1] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=31760
[2] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=31761
[3] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=31762
[4] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=702
[5] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=16942
[6] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=31764
[7] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=747
[8] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=676
[9] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=31767
[10] http://okina.univ-angers.fr/d.bonneau/publications
[11] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=31768
[12] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=31769
[13] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=31770
[14] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=31771
[15] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=31772
[16] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=31773
[17] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=31774
[18] http://okina.univ-angers.fr/publications?f%5Bauthor%5D=1071
[19] http://okina.univ-angers.fr/publications/ua18418
[20] http://dx.doi.org/10.1136/bjophthalmol-2018-312729
[21] https://bjo.bmj.com/content/early/2018/11/24/bjophthalmol-2018-312729
[22] http://www.ncbi.nlm.nih.gov/pubmed/30472657?dopt=Abstract