INTRODUCTION

Albinism is a rare genetic disorder caused by the reduction or absence of polymeric pigment melanin that affects the skin, hair, and/or eyes. Defective melanin production from tyrosine through a complex pathway of metabolic reactions leads to hypopigmentation, severe visual deficits, and finally albinism. Most patients with albinism have white hair and very light-colored skin. Skin and hair color can range from white to brown and eyes color can range from light blue to brown. Vision impairment is a major feature of all
albinism types. Several vision problems can occur, including nystagmus, iris transillumination, macular hypoplasia, strabismus, reduced visual acuity, and depth perception. There are two main albinism categories, which are classified based on the affection of skin, hair, and eyes, or only the eyes in oculocutaneous albinism (OCA) and ocular albinism (OA), respectively. In OCA, the number and structure of melanin are not significantly altered in any degree of observed pigmentation, whereas the appearance of a large number of distinct pigment cells within melanosomes is characteristic in OA. The OCA is a genetically heterogeneous and autosomal recessive disorder characterized by the hypopigmentation of skin, hair, and eyes. To date, at least seven autosomal genes have been associated with seven non-syndromic OCA (OCA1, OCA2, OCA3, OCA4, OCA6, OCA7, and OCA8), including TYR, OCA2, TYRP1, SLC45A2, SLC24A5, C10orf11, and DCT. For OCA5, the causative locus was mapped on chromosome 4q24, this is the only OCA subtype that causing genes have not been determined. Other genes included HPS1, AP3B1, HPS3, HPS4, HPS5, HPS6, DNTBP1, BLOC1S3, BLOC1S6, AP3D1, BLOC1S5, and LYST were also involved in causing two syndromic OCA: Hermansky–Pudlak syndrome (HPS1-11) and Chediak–Higashi syndrome (CHS). Syndromic OCA can be more severe and associated with additional symptoms than only alteration of pigmentation and vision. The other type of albinism is ocular albinism (OA) that affected only the eyes, caused by mutation in OA1/GPR143 gene on X chromosome. Vision acuity and photophobia of patients with OA are reduced and strabismus or nystagmus is also observed. In Vietnam, until now, the genetic data of albinism have remained unknown. Thereby, for the first time, genetic analysis was performed on seven Vietnamese albinism patients in our study.

2 | MATERIAL AND METHOD

2.1 | Subject

A total of seven affected individuals with OCA, including six (P1, P2, P3, P4, P6, and P7) and their parents (Figure 2–5) from four unrelated nonconsanguineous families, and a single man (P5) were recruited from Hanoi Medical University Hospital, Hanoi, Vietnam. All patients presented with typical clinical features of albinism, including various degrees of eyes, hair, and skin hypopigmentation. Written informed consents were obtained from all patients and family members before sample collection. This study was approved by the Institute of Genome Research Institutional Review Board, Vietnam Academy of Science and Technology.

For patients and family members, 2 ml of whole blood was collected, preserved in EDTA-containing tubes, and stored at −20°C. Genomic DNA extraction was performed by using Exgene™ Blood SV (GeneAll Biotechnology), following the manufacturer’s guidelines. Qubit™ dsDNA HS Assay Kit (Thermo Fisher Scientific) was used for DNA quantification.

2.2 | Method

2.2.1 | Exome analysis

The DNA library was constructed by Sure Select V6-Post (Agilent Technologies) following the manufacturer’s guidelines. The sequencing was performed by using an Illumina NovaSeq 6000 platform (Illumina) with paired reads of 150 bp. The reads were mapped to the hg19/GRCh37 human reference genome by the BWA.v0.7.12 tool, and Picard was used to mark the duplicates. Genome Analysis Toolkit (GATK) and Samtools were used to detect single nucleotide variants (SNVs) and short insertions/deletions (Indels). To exclude false positive, all variants with depth read lower than 20x were removed. Short Indels in the repeat regions and within the ten bp range from the start and end of the read were also excluded. After that, the remaining variants were filtered from the public databases comprising 1000G and gnomAD. All variants with minor allele frequency (MAF) under 0.01% were selected for further evaluation.

The variants were annotated with the ANNOVAR program. The in silico analysis was performed by SIFT, Polyphen-2, and Mutation Taster to anticipate the functional effect of missense and nonsense variants. The candidate variants were classified according to the Guideline and Standard of the American College of Medical Genetics and Genomics (ACMG).

2.2.2 | Sanger validation

The candidate variants were validated by direct Sanger sequencing in patients as well as their parents. Primers for PCR and sequencing were provided by PHUSA Biochem Company (Can Tho, Vietnam). For PCR amplification, 10 ng of total genomic DNA was used as a template in 20 μl of reaction mixture containing 1X Neb Master mix (New England Biolabs, Ipswich), 0.8 μl of each primer (10 pmole), and 8.4 μl of deionized water. The thermocycling was 95°C for 2 min, followed by 40 cycles of 95°C for 30s, 58°C for 30s, 68°C for 20s, and a final extension at 68°C for 5 min. The PCR products were purified using Multiscreen PCR 96 Filter Plate (Merck-Millipore) and sequenced by ABI Prism BigDye Terminator Cycle Sequencing Kit Version 3.1 (Applied BioSystems) on ABI 3500 Genetic Analyzer (Applied BioSystems).

3 | RESULT

3.1 | Clinical features

In all the seven patients, hypopigmented eyes, hair and skin, and photophobia were observed. The other ophthalmological findings, including nystagmus, reduced visual acuity, strabismus, foveal hypoplasia, ecchymosis, and reduced stereopsis, were different in each patient (Table 1).
In brothers of P1 (7 years old) and P2 (5 years old), the clinical manifestation included white hair and skin, and blue eyes (Figure 1A,B). The presentation of photophobia with discomfort and eye pain when exposed to light, nystagmus (increased when illness and stress), and choroidal metaplasia were detected. Myopia $-14$ dipters (D) and visual acuity of $1/10$ were identified in P1 and P2, respectively, and the problem to perceive depth was also observed in both patients (Table 1). Additionally, the younger brother showed a history of repeated pneumonia (P2).

Patients 3 (P3, 27 years old) and 4 (P4, 23 years old) were the first and the second child in a family of three children. The fair skin, light blonde hair, brown eyes (Figure 1C,D), photophobia, nystagmus, amblyopia, and decreased perception of depth were observed in both patients. The younger sister (P4) presented with strabismus in her right eye (Table 1 and Figure 1D), a history of glomerulonephritis, scaly dermatitis, and subcutaneous hemorrhage.

Patient 5 (P5) was a 63-year-old man with white skin and hair, blue eyes, and photophobia (Figure 1E). The patient had been visual acuity of $6/10$ in both eyes and reduced stereopsis. Manifestations of nystagmus and foveal hypoplasia were not observed in this patient (Table 1).

Patient 6 (P6) was a 26-year-old woman, the second child in the family. Clinical features of OCA consist of pinkish-white skin, hair and eyebrows, light brown eyelashes, brown eyes, and manifestations of photophobia (patient's images were not provided). The patient had normal physical, mental, and motor development, with no manifestations of nystagmus, foveal hypoplasia, and refractive errors (Table 1).

Patient 7 (P7) was a 5-year-old boy, who presented with white skin, reddish yellow to brown hair, blue eyes (Figure 1F), increased sensitivity to light (photophobia), astigmatism, and must wear glasses. Other manifestations, including nystagmus and reduced depth perception, were observed in the patient (Table 1).

### 3.2 Genetic analysis

WES data analysis revealed six candidate pathogenic variants with very rare or unknown allele frequency, including four variants in the TYR gene (NM_000372.3, #MIM:606993) (P1, P2, P3, P4, and P5), two variants in OCA2 (NM_000275.3, #MIM:611409) (P6), and HPS1 (NM_000195, #MIM:604982) (P7) genes, respectively (Table 2).

A TYR compound heterozygous mutation (c.346C>T and c.929insC) that resulted in two premature termination codons (PTCs) (p.R116* and p.R311fs*7) was observed in two brothers (P1 and P2). Both mutations were reported in dbSNP (rs61753256 and rs281865527) and were known as pathogenicity in ClinVar (Table 2). Sanger sequencing confirmed that the affected individuals inherited the c.3416C>T from the father and the c.929insC from the mother (Figure 2A). In addition, genetic testing detected c.929insC heterozygous mutation from the amniotic fluid sample of the third pregnancy of the mother (Figure 2A). Two homozygous TYR variations were found in the patients P3 and his younger

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**Table 1.** Patient characteristics

| Patient | Dermatological findings | Iris color | Photophobia | Visual acuity problem | Nystagmus | Strabismus | Foveal hypoplasia | Reduced stereopsis | Ecchymosis | Other manifestations |
|---------|-------------------------|------------|-------------|----------------------|-----------|-----------|-------------------|-------------------|------------|---------------------|
| 1       | White skin, fair hair   | Blue       | +           | Myopia of 14         | +         |           |                   |                   |            |                     |
| 2       | White skin, blond hair  | Blue       | +           | Myopia 1/10          | +         |           |                   |                   |            |                     |
| 3       | Pinkish-white skin, blond hair | Brown | +           | Amblyopia            | +         |           |                   |                   |            |                     |
| 4       | Pinkish-white skin, blond hair | Brown | +           | Amblyopia            | +         |           |                   |                   |            |                     |
| 5       | Pinkish-white skin, white hair | Brown | +           | Myopia 6/10 (-0.5D)  | +         |           |                   |                   |            |                     |
| 6       | Pinkish-white skin, light brown hair | Brown | +           | None                 | +         |           |                   |                   |            |                     |
| 7       | White skin, brown hair  | Blue       | +           | Amblyopia            | +         |           |                   |                   |            |                     |

(+) Positive; (-) Negative; (·) Not available.
sister P4 (c.115 T > C, p.W39R; Figure 2B) and P5 (c.559_560ins25, p.G190fs*12; Figure 2C). In which, the c.115 T > C has been reported in the human genome mutation database (HGMD: CM100987) and was not found in dbSNP as ClinVar. This variant resulted in a substitution of conserved amino acid tryptophan by arginine (p.W39R) and was predicted as probably damaging/deleterious by SIFT, Polyphen2, and disease causing in MutationTaster.

The c.559_560ins25 was not identified in any online databases (Table 2), but three carriers with OCA were reported in previous publications (PMID: 25577957, 31,196,117 and 31,077,556). Verification by directed sequencing showed that the heterozygous c.115 T > C variant was observed in the parent of P3 and P4 and did not inherit to the third child (Figure 2B). In brief, OCA1 subtype diagnosis was considered in five affected individuals with the finding of TYR mutations.

The other non-syndromic OCA subtype with a molecular diagnosis of OCA2 (P6) was identified to be homozygous for OCA2 c.2323G > A (dbSNP: rs774822330), leading to a substitution of glycine to serine at codon 775 in polypeptide (p.G775S) (Table 2). This missense variant was predicted to be damaging effect on protein function by SIFT, Polyphen2, and disease causing by MutationTaster. Sequencing analysis indicated that both father and mother were heterozygous carriers for c.2323G > A (Figure 3).

Only patient 7 (P7) was classified as the subtype of syndromic albinism due to the fact that the c.972delC homozygote of the HPS1 gene was found to cause Hermansky–Pudlak syndrome. The c.972delC was pathogenic mutation and was reported in dbSNP as ClinVar (Table 2). The familial segregation was confirmed for c.972delC with the heterozygous trait in both parents by Sanger sequencing (Figure 4).

4 | DISCUSSION

In this study, the molecular diagnosis for seven affected individuals with albinism was provided, including identified mutations in TYR, OCA2, and HPS1 genes associated with OCA subtype 1 (P1, P2, P3, P4, and P5), subtype 2 (P6), and Hermansky–Pudlak syndrome (HPS) (P7), respectively. OCA1 and OCA2 were the common subtypes of non-syndromic OCA, while HPS was the rare syndromic OCA.

The TYR mutations causing to OCA1 included compound heterozygote (c.346C > T and c.929insC), two homozygous of c.115 T > C (p.W39R), and c.559ins25 (p.G190Cfs*12), which were observed in two siblings (P1, P2, and P3, P4, P5) and the elderly patient (P5), respectively. There were two subtypes of OCA1 (OCA1A and OCA1B), of which OCA1A was produced by null mutations that resulted in absence or inactive tyrosinase. In OCA1B, the activity of tyrosinase was reduced compared with normal form, caused by leaky mutations, allowing some accumulation of melanin pigment over time. 8,9 Therefore, genetic testing would be considered as the important investigation for the accurate diagnosis of OCA1 subtypes.

In five OCA1 cases of our study, three patients (P1, P2, and P5) were classified as OCA1A, carrying PTC mutations (p.R116*; p.R311fs*7; and p.G190Cfs*12) in both alleles, leading to a complete loss of tyrosine function, thereby not producing melanin in the melanocytes. These mutations were known, in which the
### Table 2: Gene mutations in seven Vietnamese OCA subtype

| Patient | Gene      | Type of variant | Mutation | Zygosity | ACMG classification† | ClinVar/ACMG classification† |
|---------|-----------|-----------------|----------|----------|-----------------------|-------------------------------|
| TYR 1 and 2 | c.346C>T (p.R116*) | heterozygous | stopgain exon 1 | rs61753256 | 0.00002829 P | OCA1A |
| TYR 3 and 4 | c.115T>C (p.W39R) | homozygous | nonsense exon 1 | CM100987 | · | OCA1A |
| TYR 5 | c.559_560ins25 (p.G190Cfs*12) | homozygous | frameshift insertion | · | · | OCA1A |
| OCA2 6 | c.2323G>A (p.G775S) | homozygous | nonsynonymous exon 2 | rs774822330 | 0.000008028 LP† | OCA2 |
| HPS1 7 | c.972delC (p.M325fs*6) | homozygous | frameshift deletion | · | · | HPS1 |

Abbreviations: FS del, frameshift deletion; FS ins, frameshift insertion; het, heterozygous; hom, homozygous; HPS, Hermansky–Pudlak syndrome; LP, Likely pathogenic; nonsyn, non-synonymous; OCA, oculocutaneous albinism; P, pathogenic; PM, pathogenic moderate; PP, pathogenic supporting; PVS, pathogenic very strong.

†In current study; ‡Previously unknown zygosity; (·) Not available.

In our study, the P3 and P4 were classified as OCA1B, both carried a homozygous mutation c.115T>C (p.W39R), leading to decreased tyrosine activity and subsequently reduce melanin creation. This mutation was just reported in a Chinese male patient in a heterozygous state, who was initially diagnosed with OCA2 based on clinical features. However, upon genetic analysis, he was classified as OCA1B with compound heterozygous for c.115T>C (p.W39R) and c.1265G>A (p.R422Q) of the TYR gene. Thus, molecular screening has played an important role in the accurate diagnosis of the albinism subtype.

In our study, the TYR c.1113T>C (p.W39R) was the first observed in a homozygous state, finding in two patients of P3 and P4. In addition, not only the substitution at nucleotide 115 (c.115T>C, p.W39R) but two others at position 116 (c.116G>A, p.W39*) and 117 (c.117G>T, p.W39C) were also reported to functionally affect codon 39 in the polypeptide, all implicated in OCA1B. For these reasons, the TYR c.115T>C has been classified as "likely pathogenic" following the ACMG criteria (Table 2).

An OCA2 case detected in this study was patient P6, carrying the OCA2 homozygous variant c.2323G>A (p.G775S). This variant was previously found with homozygous in a Vietnamese patient, but the authors assumed that this substitution had no harmful consequence on protein. Preising et al. explained that, when conducting in silico analysis using SIFT and Polyphen-2, the results were contradictory, so c.2323G>A (p.G775S) was not considered to be the causative agent of the disease. Instead of the c.2323G>A (p.G775S), the homozygous c.1113T>C (p.G371C) was reported as the causative mutation in the study of Preising et al., according to novel splice donor site prediction by Splice Sequence Finder Server. However, until now, c.1113T>C (p.G371C) has been reported as a benign variant on the ClinVar database (VCV00193573.7). On the other hand, in current work, in silico prediction by SIFT and Polyphen-2, both showed the c.2323G>A (p.G775S) to be potentially damaging the encoded protein. In addition, at codon position 775 exist, two other alterations previously found in OCA2 patients including c.2323G>C (p.G775R) and c.2324G>A (p. G775D). The glycine at c.775 was highly conserved and located within the transmembrane 11 region in total of 12 domains on P polypeptide, encoded by the OCA2 gene. Therefore, the substitutions of G775 may possibly inhibit the folding of P-protein and lead to harmful consequences. Based on these suggestions, we predict that c.2323G>A (p.G775S) could be considered a "likely pathogenic" variant according to ACMG classification (Table 2) and should be functionally demonstrated in further studies.
Patient 7 was the only syndromic OCA case associated with Hermansky–Pudlak syndrome (HPS) caused by homozygous frame-shift $HPS1$ c.972delC mutation, making to the appearance of PTC on the polypeptide chain (p.M325fs*6), which was previously reported in causing of failure to the formation or resulting in a protein loss of function after translation. The $HPS1$ loss of function...
was established as a known mechanism of disease in autosomal recessive HPS. In fact, nucleotide C at position 972 was identified as a mutation “hotspot” of the HPS1 gene. The p.M325fs*6 was the co-product of the insertion (c.972insC) or deletion (c.972delC) at position 972 on the nucleotide sequence, which has been known to be the most common mutation in the European HPS patients. Therein, c.972delC was observed in some Puerto Rican, Mexican, Chinese and African American patients. The c.972insC was found mainly in Puerto Rican and some Japanese patients. In HPS, the classic clinical features included OCA, a prolonged bleeding due to storage pool-deficient platelets, and development of granulomatous, pulmonary fibrosis, or neutropenia in some cases. However, apart from the main OCA findings, the other features specialty of HPS was not identified in P3. For this reason, molecular tests could be needed for the specific classification of albinism subtypes.

5 | CONCLUSION

This is the first report on albinism-causing genes in Vietnam, exploring the mutational spectrum relevant to this disorder. To the best of our knowledge, we also report a rare TYR mutation (c.115T>C) with the novel zygosity being homozygous. Considering overlapped characteristics of OCA subtypes, molecular genetic analysis will substantially aid clinical diagnosis and genetic counseling of OCA.

AUTHOR CONTRIBUTIONS

Conceptualization: NDT, NVH; Funding acquisition: NDT; Data curation, Formal analysis, and Investigation: MTHT, VPN, NHH, TTBN, HTL, LTLA; Roles/Writing - original draft: MTHT, NDT; Writing - review & editing: MTHT, NHH, NDT, NVH.

FIGURE 3 Pedigree chart and electropherograms of P6 family with OCA2 mutation. Full/half black represents patient/carer individuals. Mutated/normal nucleotides were marked with red/blue arrows.

FIGURE 4 Pedigree chart and electropherograms of P7 family with HPS1 mutation. Full/half black represents patient/carer individuals. Mutated nucleotides were marked with red arrows.
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CONFLICT OF INTEREST

All authors declare that they have no conflict of interest.

DATA AVAILABILITY STATEMENT

The data used to support the findings of this study may be requested from the corresponding author.

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