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CASE REPORT

# A novel *de novo* ACTB variant (c.353A>T; p.K118M) associated with Baraitser-Winter syndrome type 1: a case report

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## ABSTRACT

**Background:** Baraitser-Winter syndrome type 1 (BRWS1) is a developmental disorder with low incidence, characterized by autosomal dominant congenital abnormalities, including intellectual disability, epilepsy, and structural brain malformations. The syndrome results from pathogenic mutations in the ACTB gene that encodes β-actin - a key cytoskeletal protein essential for cell structure and function.

**Case Presentation:** We report a patient with clinical features of BRWS1. Whole-exome sequencing identified a novel heterozygous missense mutation, c.353A>T (p.K118M), in the ACTB gene. The mutation results in the replacement of a conserved lysine with methionine at position 118. According to the American College of Medical Genetics and Genomics guidelines, it was classified as "likely pathogenic." The mutation was not present in population databases and was not reported earlier in HGMD or ClinVar. Parental DNA Sanger sequencing confirmed the variant to be of *de novo* origin.

**Conclusion:** This is the first clinical report of the ACTB p.K118M variant associated with BRWS1. The findings expand the mutational spectrum of ACTB and are consistent with a likely pathogenic classification, resulting in improved molecular diagnosis and understanding of the genetic basis of the condition.

**Keywords:** *ACTB, rare disease, Baraitser-Winter syndrome type 1, case report*.

## Introduction

Baraitser-Winter *s*yndrome *t*ype 1 (BRWS1; MIM #243310) is an autosomal dominant developmental syndrome that is characterized by the following primary phenotypic characteristics: hypertelorism, congenital ptosis, broad nasal root, and developmental delay (1–3). The syndrome is rare and is also associated with sensorineural hearing loss and neurological dysfunction (4,5). The syndrome is further characterized by the presence of joint stiffness, seizure activity, and musculoskeletal malformations, which collectively contribute to its phenotypic heterogeneity (6).

The vast majority of mutations in the ACTB gene are heterozygous missense variants that alter highly conserved residues within the protein structure, thereby disrupting the function of β-actin, and are the primary causative gene for the disease (2). β-actin, a cytoskeletal protein with universal distribution mapped on chromosome 7p22.1, plays a critical role in intracellular transport, cell motility, and cytoskeletal dynamics (2,3,5). Variants associated with BRWS1 frequently emerge in critical domains that are pivotal for actin polymerization and interaction with other cytoskeletal elements (2,6). Additionally, the clinical manifestations of BRWS1 are truly variable. These manifestations may encompass a wide spectrum, from musculoskeletal problems to epilepsy and cerebral disability (5,7). In a few instances only, congenital heart disease, renal malformations, and orofacial clefts were documented, once again showing that ACTB mutation is linked to multi-system involvement (8).

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Clinical reports underscore the variability in disease severity, which may be influenced by the specific mutation and additional genetic or environmental factors. A multidisciplinary approach is required for an accurate diagnosis and treatment of BRWS1, due to the complexity of its clinical presentation. Comprehensive genetic testing, such as whole exome sequencing (WES), has proven invaluable in identifying these mutations and providing a definitive diagnosis. In order to improve genotype-phenotype correlations and determine treatment approaches, the identification of new ACTB variations and their phenotypic effects continues to be significant. This study describes the clinical implications of a novel heterozygous c.353A>T variation within the ACTB gene associated with Baraitser-Winter syndrome type 1.

## Case Presentation

The patient first presented to the Pediatric Endocrinology Clinic at Near East University at the age of 5 years and 5 months due to short stature. At the time of presentation, the patient's weight was 17.4 kg (-0.74 SDS), height was 101 cm (-2.3 SDS), and BMI was 17.6 (0.89 SDS). The target-height SDS was 1.05, indicating that the patient was below their target height.

The patient had a normal prenatal history, and there were no significant findings in the natal history. The postnatal history revealed normal development of gross motor milestones. The newborn hearing screening performed postnatally was normal; however, the patient had delayed language and neuromotor development and was receiving special education support. There was no history of seizures up to the present. Physical examination revealed large, low-set, and prominent ears, high-arched eyebrows, slanted eyes, hypertelorism, and a broad nasal bridge (Figure 1B). Routine biochemical tests, thyroid hormones, celiac screening, IGF-1, and IGF-BP3 levels, which were conducted to investigate the etiology of short stature, were within normal limits. The bone age was determined to be 3 years at the time of presentation, which was delayed compared to the chronological age. In growth hormone stimulation tests using clonidine and L-dopa, a peak growth hormone response of 8.57 ng/ml was observed (<10 ng/ml for the clonidine/L-dopa protocol used at our center), indicating partial growth hormone deficiency. Other pituitary hormone screenings (ACTH, cortisol, prolactin) in this prepubertal patient were within normal limits. The patient's karyotype was found to be 46, XX. Following the molecular diagnosis, evaluations for organ involvement were performed. Abdominal ultrasound was normal, as were repeated hearing tests and eye examinations. The echocardiogram was normal. EEG results were within normal limits. Brain and pituitary magnetic resonance imaging (MRI) showed enlarged perivascular spaces, most prominently in the posterior parietal regions of both cerebral hemispheres. After the molecular diagnosis, the patient with partial growth hormone deficiency was placed under follow-up. The patient, with an annual growth rate of 7.5 cm/year, is currently being monitored without starting growth hormone treatment. The patient's last examination was performed at 6 years and 6 months of age. During the examination, the weight was 18.6 kg (-1.04 SDS), height was 108.5 cm (-1.99 SDS), and BMI was 15.8 (0.14 SDS). The patient is being closely monitored for growth.

Afterward, the patient was referred to the Near East University Medical Genetic Clinic for developmental issues, such as congenital ptosis. During the examination, hypertelorism and dysmorphic facial features such as a long philtrum, pear-shaped nose, and short stature were observed. According to her mum, she was born weighing 3250 g in weight and measuring 48 cm in height. She is under special education, for instance, speech and ergotherapy, because of her hyperactivity. She has one healthy brother, and her parents are not of advanced age (both 29 years old when she was born) (Figure 1A). Moreover, the proband has two paternal family members who were affected by Duchenne muscular dystrophy. This DMD family history is unrelated to the ACTB finding and did not influence the diagnostic workup for the proband's developmental phenotype; it is reported here only for completeness of the family history

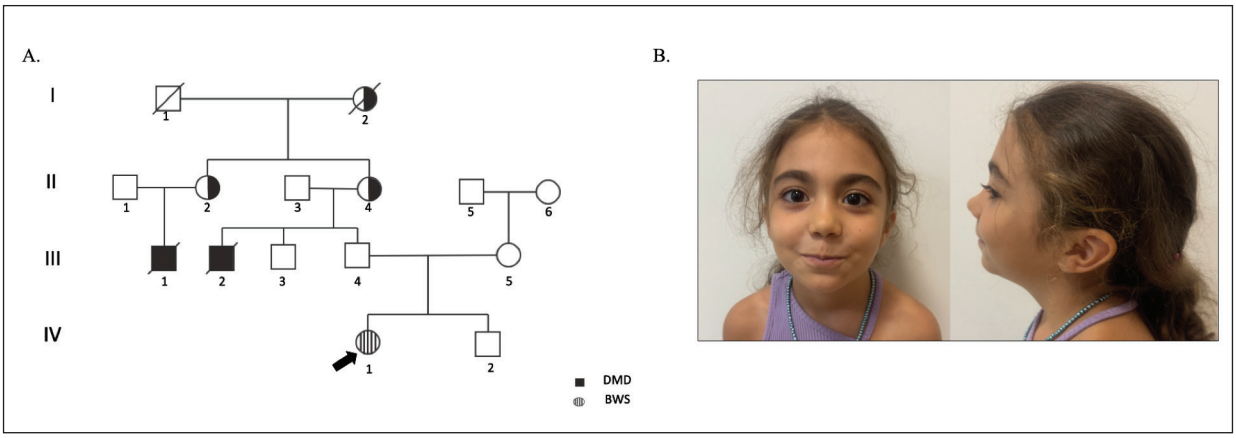



***Figure 1.*** *A – Family pedigree of the proband. The pedigree illustrates two independent conditions in this family: (i) an X-linked recessive DMD trait, with affected males (III-1, III-2, both deceased) and presumed female carriers (II-2, II-4); and (ii) an apparently isolated, de novo occurrence of BRWS1 in the proband (IV-1, indicated by an arrow, striped shading), with no evidence of transmission from either parent. B - Facial photographs of the proband.*

### *Sample collection and genetic testing workflow*

After the patient's and her parents' informed consent, venous peripheral blood samples were taken. Automatic Qiagen DNA isolation kits (Cat. No. 951034, QIAGEN, Hilden, Germany) were utilized to isolate high-quality Genomic DNA (gDNA) to ensure the delivery of genetic material according to manufacturer instructions. After quality and concentration control, gDNA was fragmented using an enzyme-based method, followed by enrichment through the use of DNA capture probes targeting approximately 41 Mb of the coding exome and the mitochondrial genome, as defined by the GRCh38/hg38 reference genome. Subsequent to this, sequencing libraries were prepared and analyzed on an Illumina platform, resulting in the attainment of >20x coverage in more than 98% of targeted regions. Variant identification, filtration, and annotation were conducted using in-house bioinformatics pipelines. Variants with a minor allele frequency below 1% in the gnomAD database and those reported in HGMD, ClinVar, or CentoMD were evaluated.

The potential pathogenicity of variants was then classified according to the guidelines set out by the American College of Medical Genetics and Genomics (ACMG) (9). Additionally, applying the ACMG/AMP framework (9), the following evidence codes were met: PM6 (*de novo* occurrence inferred from targeted Sanger analysis showing absence of the variant in both parents; independent confirmation of biological maternity and paternity was not separately performed), PM2 (absence from population databases, including gnomAD), PP3 (predicted deleterious by multiple *in silico* predictors aggregated via the VarSome ACMG classifier), and PP4 (patient phenotype specific to an ACTB-related disorder). Moreover, parental segregation analysis to confirm a *de novo* mutation was conducted by traditional Sanger sequencing using an ABI 3500 DNA sequencer, and the data obtained were analyzed against reference sequences according to the GenBank and Ensembl databases.

### *In Silico structural modelling*

The three-dimensional structure of human β-actin was obtained from the AlphaFold Protein Structure Database (UniProt accession P60709) (10,11), generated using AlphaFold2 (10). The p.K118M substitution was introduced into the wild-type model using the mutagenesis function in PyMOL (Schrödinger, LLC) (12), and the wild-type and mutant models were superimposed and visually compared to assess local conformational change. Predicted per-residue confidence (pLDDT) scores from the AlphaFold model were used to gauge the reliability of the region surrounding residue 118. Hydrogen bonding and electrostatic interaction changes were inferred in PyMOL by comparing wild-type and mutant models, focusing on residues within 5 Å of position 118 and its proximity to the nucleotide (ATP)-binding cleft. All software was used with default parameters unless otherwise specified.

## Results

Whole-exome sequencing has revealed a heterozygous *de novo* likely pathogenic c.353A>T variant within the *ACTB* gene (NM_001101.5), resulting in an amino acid substitution of a positively charged lysine with methionine at the highly conserved 118th amino acid position (p.K118M). Figure 2A shows the Integrative Genomics Viewer (IGV), aligned on chromosome 7

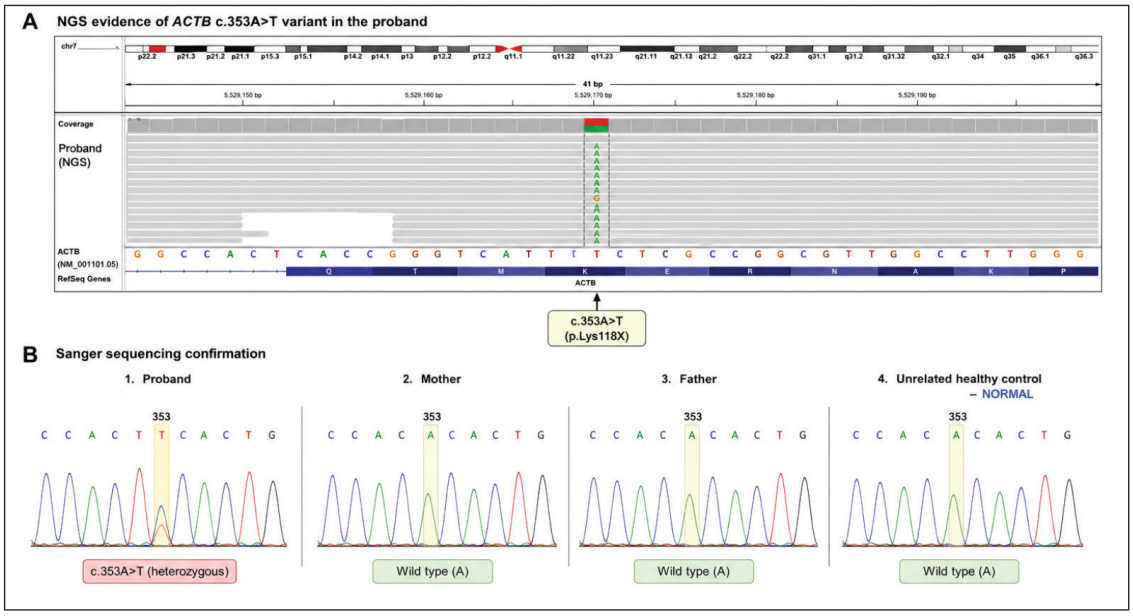



***Figure 2.*** *A - IGV visualization of sequencing reads aligned to the ACTB gene at chr7:5,529,171. B - Sanger Sequencing Analysis of the ACTB gene: Sanger sequencing chromatograms illustrating the genetic profile of the p.K118M mutation in the ACTB gene. (B2) The proband, (B2) the sequencing result from the mother, (B3) the sequencing result from the father, and (B4) the sequencing result from a healthy control. Variations in nucleotide sequences are highlighted, demonstrating the presence or absence of the mutation.*

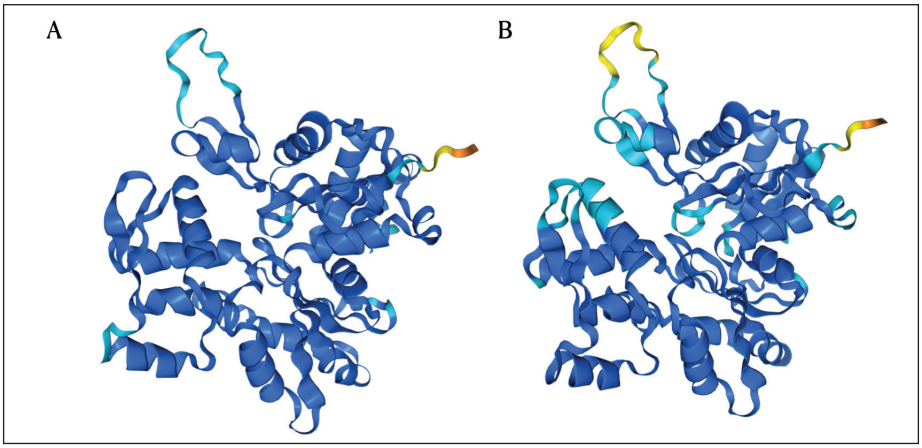



***Figure 3.*** *(A) Wild-type β-actin with lysine (K) at position 118. (B) Mutant β-actin after the p.K118M substitution. Structural comparison is illustrative; see Methods for the specific in silico analyses supporting the predicted interaction changes discussed in the text.*

(GRCh38/hg38) precisely at position 5,529,171 of the *ACTB* gene.

While the WES result identified the heterozygous missense 353A>T variant in the proband, her parents did not show the presence of the mutation. Targeted Sanger mutation analysis of the *ACTB* c.353A>T p.(Lys118Met) variant was performed in both parents. The variant was not detected in the father or in the mother, confirming absence of the variant in both parents and supporting a *de novo* origin in the proband (Figure 2B). The variant was absent from gnomAD, ClinVar, and HGMD Professional, and was not previously reported in the literature, supporting its designation as novel.

Additionally, applying the ACMG/AMP framework, the following evidence code was met: PM6. This finding is consistent with the VarSome ACMG-based classification of the variant as likely pathogenic. The variant's *de novo* occurrence and absence from population databases support, but do not alone establish, its pathogenic role in the patient's clinical presentation; this classification is interpreted as likely pathogenic pending functional confirmation (14, 15).

## Structural impact of the p.K118M mutation in β-Actin

The p.K118M substitution replaces lysine (K), a basic and hydrophilic residue, with methionine (M), a neutral, nonpolar, and hydrophobic residue, at position 118 of β-actin - a substantial physicochemical change (Figure 3). Structural modeling using the AlphaFold-predicted human β-actin structure (Methods, "*In Silico* Structural Modeling") showed high per-residue confidence (pLDDT) in the region surrounding position 118, supporting the reliability of this region of the model. Superposition of the wild-type and p.K118M mutant models in PyMOL indicated changes in predicted hydrogen-bonding and electrostatic interactions involving residues within 5 Å of position 118, a region located close to the nucleotide (ATP)-binding cleft of β-actin. These *in silico* findings suggest that the p.K118M substitution may locally alter the electrostatic and hydrogen-bonding environment near the ATP-binding region of β-actin; experimental studies are required to determine whether this substitution has a functional effect on ATP binding, filament stability, or polymerization.

## Discussion

Identification of a heterozygous c.353A>T (p.K118M) variant in the *ACTB* gene in a patient with Baraitser-Winter syndrome type 1 (BRWS1, MIM: #243310) is more evidence for the pathogenicity of the β-actin mutation (5). Notably, the mutation is not present in the HGMD Professional or ClinVar databases and is therefore described as novel (9). The mutation is defined by substitution of methionine (M) at a conserved position, rather than lysine (K), which is characteristically observed in the wild-type sequence. As per the recommendations provided by the ACMG, the mutation was designated as "likely pathogenic," and the clinical significance thereof was well-supported

(9). The *ACTB* gene that codes for β-actin is a core component of the cytoskeleton and is vital in the process of supporting cellular structure, intracellular transport, and coordinating several processes of signaling necessary for optimal functioning of the cell (16). Given the central role of actin polymerization and depolymerization reactions in regulating cytoskeletal dynamics, it is unsurprising that mutations that disrupt these reactions can be detrimental to development and function (6,8,18).

The p.K118M mutation, as defined in the present study, is the replacement of a positively charged lysine residue by methionine at a very conserved position close to the ATP-binding site of β-actin. This alteration has been shown to have a profound effect on the physicochemical characteristics of the protein that may lead to decreased filament stability, misfolding, and disrupted cytoskeletal assembly (18,19). Moreover, computational structural modelling shows that such substitution may lead to disruption of hydrogen bonding and electrostatics and an increase in the tendency for misfolding and aggregation, features of actinopathy-linked cytoskeletal dysfunction (23).

Clinically, BRWS1 is characterized by a spectrum of congenital abnormalities including intellectual disability, epilepsy, craniofacial dysmorphism, and structural brain anomalies (1,17). Classification of this patient as BRWS1 was based on the presence of core diagnostic features established for this syndrome —- hypertelorism, congenital ptosis, a broad nasal bridge, and developmental delay (1( - which together are considered sufficiently specific for the diagnosis even in the absence of every reported feature. Notably, this patient lacks a history of seizures and lacks a classical major structural brain malformation, both of which are recognized, though not obligatory, features of BRWS1 (1,8). This does not exclude the diagnosis, as considerable phenotypic heterogeneity has been documented among ACTB-related disorders, with no single feature being present in all reported cases (8, 22). Nonetheless, we acknowledge that in the absence of seizures and major structural brain malformation, this patient's presentation could alternatively be considered within the broader, variable ACTB-related disorder spectrum rather than strictly typical BRWS1, and we present the classification here as the best-fitting diagnosis based on the core features observed (24).

Molecular diagnosis was significant in this case since it confirmed the *de novo* nature of the *ACTB* c.353A>T variant (13,14). This was confirmed by a lack of mutation in both parents, ruling out its presence within the population databases. The variant was "likely pathogenic" according to the ACMG criteria, thus supporting its likely pathogenic classification (9). Despite its lack of occurrence in HGMD and ClinVar, pathogenicity for the variant has been warranted in consideration of established β-actin function to preserve cellular homeostasis and is also evidenced by the impact of missense mutations in regions crucial to function (5).

ACTB mutations have been found to cause aberrant protein-protein interactions with essential cytoskeletal regulators, cofilin and profilin, that increase cytoskeletal defects (20). Of particular interest is the p.K118M mutation, which is near the region of the ATP-binding site, as it not only affects binding and hydrolysis of ATP but also causes defective polymerization of actin, decreases filament stability, and produces abnormally assembled cytoskeletons (19, 21). These molecular defects cause the disruption of vital cellular functions, including intracellular trafficking, signal transduction, and embryonic neural crest cell migration, and thus type 1 Baraitser-Winter syndrome craniofacial and musculoskeletal dysmorphisms (5). Besides, conserved actin residues have been fully described as being toxic, owing to β-actin malfunction being linked to neurological impairment, epilepsy, stiff joints, and organ insufficiency (1, 23). As β-actin also interacts with several cytoskeleton regulators, the mutation p.K118M will disrupt the actin filaments and thereby may further decrease cytoskeleton strength and continuously contribute to BRWS1 disease pathology (23, 24). Further experimental and computational research is required to fully shed light on the molecular and functional consequences of this mutation (25).

Genetic counselling of the affected kindred is essential after pathogenic designation of the *ACTB* c.353A>T mutation. Full disclosure of recurrence risk, potential impact on other members of the kindred, and therapeutic interventions provided should be communicated. Given the confirmed *de novo* occurrence, the empiric recurrence risk for the parents in future pregnancies is low but not zero, owing to the possibility of parental germline mosaicism; this is distinct from the approximately 50% risk of transmission the patient herself will face in future pregnancies as an affected individual. Both risk figures should be communicated during genetic counselling. Future *in vitro* studies through actin polymerization assays and co-immunoprecipitation with proteins controlling the cytoskeleton should provide more insight into the functional impact of the mutation. Furthermore, patient-derived fibroblast models could also offer informative perspectives on cellular morphology and actin filament organisation in BRWS1. Such future work would also have to evaluate the expansion of patient registries for the purpose of improving genotype-phenotype correlations to become more accurate and improve BRWS1 and other *ACTB* disorder clinical management planning.

In summary, the present study contributes to the previous knowledge regarding the *ACTB* mutation and developmental disorders. Identification of this new variant helps to expand the spectrum of BRWS1 mutations, thus emphasizing the necessity of comprehensive genomic screening in patients with complicated congenital disorders.

## Conclusion

We report a novel, *de novo ACTB* variant, c.353A>T (p.K118M), classified as likely pathogenic, in a patient with a phenotype compatible with an *ACTB*-related developmental disorder. *In silico* modeling suggests the substitution may affect β-actin structure and stability, but functional studies are required to establish the molecular consequences of p.K118M. Molecular diagnosis and multidisciplinary follow-up remain important for patient care and for identifying potential complications of *ACTB*-related disorders. Further research into *ACTB* variants will help clarify genotype-phenotype correlations and, in time, may inform targeted therapeutic approaches.

### Statement of Ethics

This research was conducted ethically according to the World Medical Association Declaration of Helsinki. Informed consent for genetic testing was obtained from the family according to national regulations on ethics. Informed consent was obtained in writing from all participants or guardians of the concerned participants for publishing this case report, including specifically for publication of identifiable facial photographs (Figure 1B).

### Declaration of conflicting interest

The authors of this article have no affiliations with or involvement in any organization or entity with any financial interest or nonfinancial interest in the subject matter or materials discussed in this manuscript.

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None.

**Author contributions**

Conceptualization, B.K., A.T., D.D., and M.Ç.E.; Diagnosis, and sample collection, N.A.; Resources, M.Ç.E.; Investigation and conducting experiments, B.K., D.D., A.T. & M.C.E.; Formal analysis, M.C.E.; Visualization, B.K., N.A., and M.Ç.E.; Writing - original draft preparation, B.K.; Review, M.C.E.; Writing - review and editing, B.K., A.T., D.D., N.A., M.C.E..; Supervision, M.C.E.; Project administration, M.C.E.; All authors have read and agreed to the published version of the manuscript.

**Data availability statement**

Data are available upon request.

**Ethical approval**

This case report was formally exempt from ethics committee approval in accordance with applicable institutional requirements. Written informed consent was obtained from the patient's legal guardian for genetic testing and publication of the case report, including identifiable facial photographs.

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