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Autism Spectrum Disorder in a Boy with Clinically Diagnosed Kabuki Syndrome and a YWHAG Variant of Uncertain Significance: A Case Report

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Abstract **Case Report**

Background: Kabuki syndrome is a rare multisystem neurodevelopmental disorder usually caused by pathogenic *KMT2D* or *KDM6A* variants, although a substantial minority of clinically typical patients remain molecularly unresolved. Autism spectrum disorder (ASD) has been reported in this population, but its prevalence and relationship to genotype remain poorly characterized. ***Case Presentation:*** We report a 12-year-old Moroccan boy, born to consanguineous parents, referred for severe behavioral disturbances including psychomotor hyperactivity, self-injurious behavior, and marked language delay. Physical examination showed characteristic craniofacial dysmorphism, long palpebral fissures with eversion of the lateral lower eyelids, blue sclerae, broad arched eyebrows, a depressed nasal tip, and a high-arched palate, together with microcephaly, postnatal growth restriction, infantile hypotonia, a waddling gait, and possible scoliosis, fulfilling international consensus criteria on dysmorphology, growth restriction, hypotonia, and musculoskeletal abnormalities for a clinical diagnosis of Kabuki syndrome. He also presented with severe ASD, attention-deficit/hyperactivity disorder, intellectual developmental disorder, and generalized epilepsy, established according to DSM5 criteria. Whole-exome sequencing identified no pathogenic variant in *KMT2D* or *KDM6A*, but revealed a heterozygous variant of uncertain significance in *YWHAG* (NM_012479.3:c.340G>A; p.Glu114Lys), a gene associated with developmental and epileptic encephalopathy. Pharmacological management included extended-release methylphenidate, which was discontinued because of paradoxical worsening of hyperactivity, followed by low-dose aripiprazole, which improved self-injurious behavior and agitation but caused a persistent tremor. ***Conclusions:*** This case shows that a well-characterized clinical gestalt remains sufficient to diagnose Kabuki syndrome despite negative first-line genetic testing, and that residual possibilities, including deep intronic, structural, or regulatory variants, and multilocus ("blended") phenotypes, should be considered before further investigation is abandoned. The co-occurring *YWHAG* variant cannot presently be assigned a causal role but merits targeted follow-up. Recognition of ASD and ADHD as distinct comorbidities had important implications for individualized behavioral and pharmacological management, and systematic psychiatric assessment should be integral, not incidental, to the multidisciplinary evaluation of Kabuki syndrome.

Keywords: Kabuki syndrome; autism spectrum disorder; YWHAG; variant of uncertain significance; whole-exome sequencing; blended phenotype; case report.

INTRODUCTION

The psychiatric phenotype of Kabuki syndrome remains under-recognized despite being a major determinant of long-term functioning, adaptive outcome, and family burden. Kabuki syndrome (KS; OMIM #147920) is a rare multisystem neurodevelopmental disorder characterized by a distinctive pattern of craniofacial dysmorphism, developmental delay, intellectual disability, postnatal growth impairment, and multiple congenital anomalies [1,2]. Although pathogenic variants in *KMT2D* and, less frequently, *KDM6A* account for most molecularly confirmed cases, the syndrome exhibits considerable clinical and genetic heterogeneity, and a proportion of patients fulfilling accepted clinical diagnostic criteria remain without an identifiable molecular cause despite comprehensive genetic testing [2,3,4,5,11]. Consequently, the diagnosis of Kabuki syndrome continues to rely on careful clinical phenotyping in genetically unresolved cases [3].

Beyond its dysmorphic and congenital manifestations, Kabuki syndrome is increasingly recognized as a complex neurodevelopmental disorder with a broad behavioral phenotype. Intellectual disability, language impairment, attention-deficit/hyperactivity disorder (ADHD), anxiety, sleep disturbances, emotional dysregulation, and autistic features have all been described, yet the prevalence and clinical significance of autism spectrum disorder (ASD) remain uncertain because available evidence is largely limited to case reports and small observational cohorts [6–10]. This phenotypic variability suggests that additional genetic or biological factors may influence disease expression in some individuals.

The increasing use of whole-exome sequencing (WES) has also revealed that a subset of patients with rare genetic disorders harbor multiple potentially relevant genomic findings. Such multilocus diagnoses or blended phenotypes may contribute to unexpectedly severe or atypical clinical presentations and represent an emerging challenge for genotype–phenotype interpretation [15,16]. In parallel, variants in

YWHAG, a gene encoding the 14-3-3γ protein involved in neuronal development and synaptic signaling, have been associated with developmental and epileptic encephalopathy accompanied by developmental delay, epilepsy, intellectual disability, and autistic features, resulting in substantial phenotypic overlap with several neurodevelopmental syndromes [13,14].

Here, we report a boy with clinically diagnosed Kabuki syndrome who presented with severe ASD,

ADHD, generalized epilepsy, and intellectual developmental disorder despite the absence of pathogenic *KMT2D* or *KDM6A* variants. Whole-exome sequencing identified a heterozygous *YWHAG* variant of uncertain significance. This case highlights the diagnostic challenges posed by genetically unresolved Kabuki syndrome, explores the potential contribution of multilocus genetic mechanisms to phenotypic severity, and underscores the importance of integrating detailed psychiatric phenotyping with genomic investigation in rare neurodevelopmental disorders.

CASE PRESENTATION

Patient Information

A 12-year-old Moroccan boy was first referred to our child and adolescent psychiatry department in 2023 for severe behavioral disturbances characterized by psychomotor hyperactivity, recurrent temper tantrums, self-injurious behavior, impaired social interaction, and marked language delay.

He was the second of two living children and attended primary school. The patient was born to consanguineous parents, reported as related in the first degree; the exact nature of this relationship could not be further specified. His mother was 31 years old, gravida 4, para 2. Her first pregnancy ended in spontaneous abortion at two months of gestation; her second pregnancy resulted in the patient's older brother, now aged 17 years, who had reportedly achieved normal psychomotor development; her third pregnancy was the index patient; and her fourth pregnancy ended in fetal loss during the second trimester. His father was 44 years old, retired, and had no relevant medical history.

Prenatal, Perinatal, and Early Developmental History

The patient was born following an adequately monitored, uncomplicated pregnancy. Delivery occurred at term by medically assisted vaginal birth. His birth weight was 3,000 g. A delayed cry after birth was reported, although no additional neonatal resuscitation data were available.

His development was globally delayed. He achieved independent sitting at approximately 12 months and walking at four years of age. His first words emerged at approximately two years of age, followed by reported language regression at three years. Language development subsequently remained markedly impaired. Toilet training had not been acquired by the time of assessment. His vaccinations were administered according to the Moroccan National Immunization Program.

Medical and Neurological History

The patient developed a febrile episode on the second day of life, which was treated on an outpatient basis with paracetamol. At four months of age, he was hospitalized for two weeks because of fever associated with febrile status epilepticus. He was hospitalized again at one year of age for an undocumented febrile illness.

Since 2019, he had been followed by pediatric neurology with a previous diagnosis of cerebral palsy, initially attributed to presumed neonatal distress, together with generalized epilepsy. Treatment included sodium valproate and clobazam. Brain magnetic resonance imaging showed no structural abnormality.

Electroencephalograms performed in 2019 and 2021 were compatible with idiopathic generalized epilepsy. A further electroencephalogram in 2024 showed generalized epileptiform activity considered probably genetic in origin.

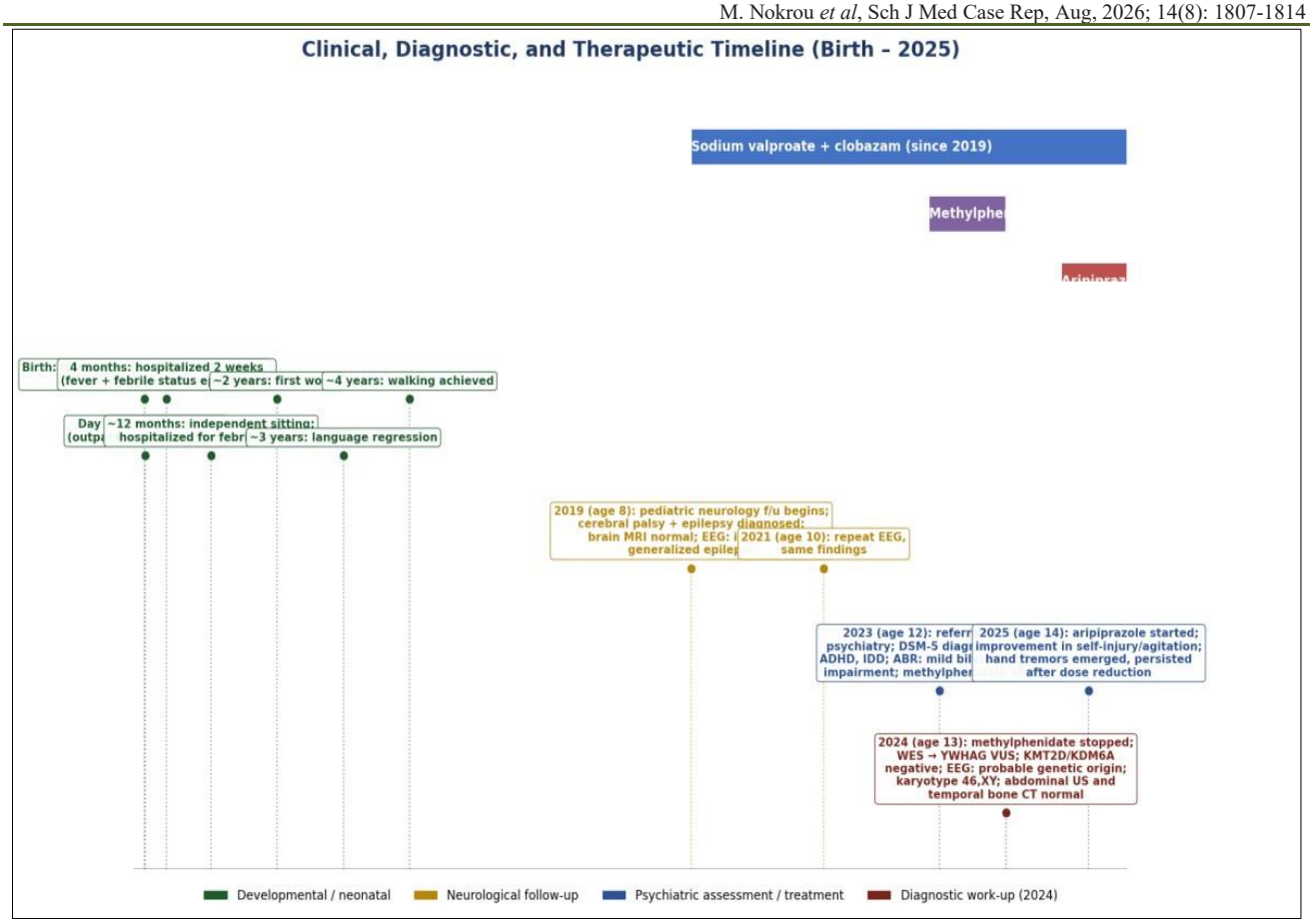

Figure 1: Clinical, Diagnostic, and Therapeutic Timeline

Top panel: duration of pharmacological treatments. Bottom panel: key developmental, neurological, psychiatric, and diagnostic milestones from birth to 2025. BW: birth weight; f/u: follow-up; ABR: auditory brainstem response; WES: whole-exome sequencing; VUS: variant of uncertain significance.

Neurodevelopmental and Psychiatric History

According to his mother, the patient's neurodevelopmental difficulties had been apparent from the first years of life. He did not establish adequate eye contact, display a social smile, point to share interest, or engage in joint attention. He showed limited social reciprocity, did not spontaneously share pleasure or interests with others, and had marked deficits in both verbal and nonverbal social communication. He showed little interest in interacting or playing with peers.

Restricted and repetitive behaviors included motor and vocal stereotypies. These manifestations occurred in the context of global developmental delay, severe language impairment, marked hyperactivity, temper tantrums, and self-injurious behavior.

During psychiatric examination, the patient was markedly restless and displayed repetitive motor and vocal behaviors. Contact was difficult to establish. He did not consistently respond when his name was called and did not maintain eye contact. His expressive language was poor and qualitatively impaired.

On the basis of the developmental history and clinical psychiatric assessment, diagnoses of autism spectrum disorder and attention-deficit/hyperactivity disorder were retained according to DSM-5 criteria. No standardized autism assessment instrument, such as the ADOS-2 or ADI-R, was administered. A clinical diagnosis of intellectual developmental disorder was also retained on the basis of the marked global developmental delay and severe adaptive impairment; no standardized cognitive or adaptive functioning assessment was available.

Physical Examination and Dysmorphology

At the initial evaluation, at age 12 years, the patient was conscious and clinically stable. His height was 135 cm and his weight was 24 kg, indicating marked postnatal growth restriction. Physical examination showed a characteristic facial dysmorphism, microcephaly, hypersalivation, and a waddling gait. Deep tendon reflexes were present and symmetrical.

The dysmorphic findings included long palpebral fissures with eversion of the lateral third of the lower eyelids, blue sclerae, broad arched eyebrows with sparse lateral portions, a depressed nasal tip, a short nasal columella, a trapezoid-shaped philtrum, large prominent ears, and a high-arched palate.

Additional clinical manifestations compatible with Kabuki syndrome included possible scoliosis, hypotonia, postnatal growth restriction, developmental delay, intellectual developmental disorder, and generalized epilepsy. A previous history of aspiration pneumonia was also reported.

The combination of developmental delay or intellectual disability, infantile hypotonia, and characteristic facial features supported a clinical diagnosis of Kabuki syndrome despite the absence of an identified pathogenic variant in KMT2D or KDM6A.

Table 1. Clinical Features Compatible with Kabuki Syndrome Identified in the Patient

Domain	Findings in this patient
Craniofacial dysmorphism	Long palpebral fissures with eversion of the lateral lower eyelid; blue sclerae; broad, arched eyebrows with sparse lateral portion; depressed nasal tip; short columella; trapezoid-shaped philtrum; large prominent ears; high-arched palate; microcephaly
Skeletal	Possible scoliosis
Growth	Postnatal growth restriction (height 135 cm and weight 24 kg at age 12 years)
Neurological / neuromuscular	Hypotonia; waddling gait; hypersalivation; seizures (generalized epilepsy)
Cognitive / developmental	Global developmental delay; intellectual developmental disorder
Other	Mild bilateral hearing impairment, more pronounced in the left ear; history of aspiration pneumonia

No standardized clinical scoring instrument (e.g., Niikawa-Kuroki score) was formally applied; findings are grouped here by clinical domain for the reader's reference. Cardiovascular assessment (see Complementary Investigations) was unremarkable.

Complementary Investigations

Cardiological assessment was unremarkable. Color Doppler echocardiography showed normal cardiac anatomy, preserved biventricular function, and no valvular abnormality, intracardiac shunt, or obstruction of the right or left ventricular outflow tract.

Abdominal ultrasonography performed in 2024 showed no abnormality. Temporal bone computed tomography performed in 2024 was also normal.

Auditory brainstem response testing in 2023 showed an auditory threshold of 30 dB in the right ear and 40 dB in the left ear, suggesting mild bilateral hearing impairment, which was more pronounced on the left.

Constitutional karyotyping showed a normal male karyotype, 46, XY, without a detectable chromosomal abnormality. Serum immunoglobulin A, G, and M concentrations were within normal limits.

Molecular Genetic Findings

Whole-exome sequencing was performed in 2024. No pathogenic or likely pathogenic variants were identified in KMT2D or KDM6A.

A heterozygous missense variant was identified in exon 2 of the YWHAG gene:

NM_012479.3(YWHAG):c.340G>A, predicted to result in the amino acid substitution p.(Glu114Lys). The variant was present in approximately 50.48% of sequencing reads, consistent with a heterozygous state. The substitution of glutamic acid by lysine at position 114 was noted by the laboratory to represent a relatively conservative physicochemical change.

The laboratory reported that this variant had been observed in the gnomAD reference population at a very low frequency of approximately 0.0002%. At the time of analysis, it had not been reported in ClinVar or in the published scientific literature. In-silico prediction algorithms yielded conflicting results, and no published functional study was available to establish its biological effect. The variant was therefore classified as a variant of uncertain significance (VUS).

Pathogenic variants in YWHAG have been associated with autosomal dominant, nonspecific epileptic encephalopathy, a condition that may present with epilepsy, psychomotor delay, autistic features, and hyperactivity. The phenotypic overlap raised the possibility that the YWHAG variant might contribute to the patient's neurological and behavioral manifestations. Nevertheless, because it remained a variant of uncertain significance, no causal relationship could be established.

Diagnostic Synthesis

Taken together, the clinical evaluation supported three concurrent diagnostic conclusions: (1) autism spectrum disorder and attention-deficit/hyperactivity disorder according to DSM-5 criteria, together with a clinically diagnosed intellectual developmental disorder in the absence of standardized cognitive testing; (2) clinically diagnosed Kabuki syndrome (OMIM #147920), based on the characteristic combination of developmental delay, infantile hypotonia, postnatal growth restriction, skeletal abnormalities, and typical facial features, despite the absence of a pathogenic or likely pathogenic variant in KMT2D or KDM6A; and (3) a heterozygous variant of uncertain significance in YWHAG, a gene associated with epilepsy and neurodevelopmental disorders, whose

contribution to the patient's phenotype remains uncertain.

Therapeutic Interventions and Follow-up

The patient received speech and language therapy and psychomotor rehabilitation. He continued neurological follow-up and treatment with sodium valproate and clobazam for generalized epilepsy.

In 2023, extended-release methylphenidate was initiated at a dose of 18 mg once daily for severe hyperactivity and attention difficulties. The exact treatment duration and adherence could not be determined. In 2024, the patient's mother returned because of worsening hyperactivity, leading to methylphenidate discontinuation.

In 2025, aripiprazole was introduced at 2.5 mg daily. Treatment was associated with an improvement in self-injurious behavior and psychomotor agitation, but no clear improvement in attention or concentration was observed. The patient subsequently developed prominent hand tremors. The aripiprazole dose was reduced to 1.25 mg daily; however, the tremors persisted despite dose reduction. Further clinical evolution, including any decision regarding continuation or discontinuation of aripiprazole, was not available at the time of manuscript preparation.

Patient Perspective

Given the patient's age and the severity of his neurodevelopmental and communication impairments, a direct first-person account of his experience could not be obtained. His mother served as the primary informant throughout the diagnostic and therapeutic process and consistently reported on his behavioral difficulties, response to treatment, and functional status at each stage of follow-up.

Consent

Written informed consent for publication of the clinical information was obtained from the patient's legal guardian. No identifiable clinical photographs will be published.

DISCUSSION

Why was this phenotype unusually severe?

This case is defined less by an isolated diagnosis of Kabuki syndrome than by the convergence of features rarely reported together: a clinical Kabuki phenotype without a *KMT2D*/*KDM6A* variant, a *YWHAG* variant of uncertain significance, severe autism spectrum disorder (ASD), attentiondeficit/hyperactivity disorder (ADHD), and generalized epilepsy. The central question this observation raises is why the phenotype was unusually severe, and whether more than one genetic mechanism contributed to it.

A clinical diagnosis of Kabuki syndrome

Kabuki syndrome (OMIM #147920) [1] is caused by pathogenic *KMT2D* or *KDM6A* variants in most molecularly confirmed cases [2,3,11], though a substantial minority of clinically typical patients remain unresolved, and international criteria accept a clinical diagnosis based on the characteristic facial gestalt, hypotonia, developmental delay, and musculoskeletal findings [2,4]. Our patient exhibited the characteristic craniofacial gestalt, postnatal growth restriction, infantile hypotonia, generalized epilepsy, and additional supportive clinical findings consistent with Kabuki syndrome, fulfilling the international consensus diagnostic criteria.

Three elements support this diagnosis with reasonable confidence: a facial gestalt that was highly specific rather than merely suggestive, full rather than partial satisfaction of published consensus criteria [4], and exclusion of the principal differential, CHARGE syndrome, based on normal cardiac, renal, and temporal bone imaging. Certainty ultimately requires molecular or epigenetic confirmation.

Why might sequencing have been negative?

A negative *KMT2D*/*KDM6A* result on whole-exome sequencing does not equate to a negative genetic evaluation. Deep intronic, structural, and regulatory variants, as well as mosaicism, are not reliably captured by exome sequencing and have been implicated in unsolved Kabuki syndrome [3,5]. Reanalysis, whole-genome sequencing, or genome-wide methylation profiling remain plausible next steps for this patient before a genetic explanation is abandoned.

Autism spectrum disorder in Kabuki syndrome

The prevalence of ASD in Kabuki syndrome is unknown. Evidence is limited to case reports and small series: Sari *et al.,*[6] and Sertçelik *et al.,*[8] each described a single child meeting ASD criterion; Parisi *et al.,*reported three children, without prevalence data [7]. Larger neurobehavioral cohorts document substantial heterogeneity in language, motor, and adaptive function [9] and a high burden of anxiety [10], but neither was designed to estimate ASD prevalence.

Diagnosing ASD against a background of moderate-to-severe intellectual disability is notoriously difficult, because global cognitive delay can itself produce reduced eye contact, limited reciprocal communication, and repetitive behavior. ASD should therefore be diagnosed only when social communication deficits and repetitive patterns exceed what the child's overall developmental level would predict [2,4], a distinction that carries real clinical consequences: it determines whether intervention targets autism-specific social-communication goals or broader developmental stimulation alone. A multidisciplinary assessment, combining child psychiatry, developmental pediatrics, and speech language pathology, ideally with

standardized instruments such as the ADOS-2, is therefore advisable whenever ASD is suspected in a syndromic, intellectually disabled child.

Our patient met DSM-5 ASD criteria: absent social smiling and joint attention, poor eye contact, motor and vocal stereotypies, and inconsistent response to name, in a pattern closely resembling the case of Sertçelik *et al.,*[8]. No standardized instrument (ADOS-2, ADI-R) was used, a limitation shared by most published Kabuki–ASD cases. Clinically, however, the ASD diagnosis was not merely descriptive: it explained his profound difficulty engaging with structured rehabilitation and verbal instruction during speech therapy, and the social withdrawal that compounded his family's caregiving burden. Recognizing ASD as a distinct, additional diagnosis, rather than attributing his entire presentation to intellectual disability, justified targeted behavioral strategies rather than generic developmental stimulation alone.

YWHAG: modifier, coincidence, or second diagnosis?

Whole-exome sequencing identified a heterozygous VUS in *YWHAG*, encoding the 14-3-3γ protein. Pathogenic *YWHAG* variants cause developmental and epileptic encephalopathy 56, with epilepsy, developmental delay, intellectual disability, and autistic or hyperactive features [13,14], a profile overlapping substantially with our patient's epilepsy, ADHD, and ASD.

Multiple molecular diagnoses occur in approximately 5–8% of exome-solved rare-disease cases, more often in consanguineous families [15,16], and are termed “blended” when both conditions produce overlapping features [16]. This has been directly documented in Kabuki syndrome: one *KMT2D*-positive patient with an additional 12p13.31 deletion showed a substantially more severe phenotype than other patients in the same series, attributed specifically to the second genomic finding [17]. This precedent supports a testable hypothesis for our patient: an unidentified *KMT2D*/*KDM6A* variant, alongside the *YWHAG* VUS, could jointly explain both the classic gestalt and the disproportionate severity observed here.

Although biologically plausible, this hypothesis remains speculative. Current ACMG/AMP guidance is explicit that a VUS should not be used to explain a phenotype without segregation or functional evidence [18], and parental testing, not performed here, would meaningfully clarify its status. Until such data exist, *YWHAG* is best regarded as a candidate modifier, not an established second diagnosis.

Psychiatric comorbidity and its clinical assessment

ADHD symptoms, attention difficulties, impulsivity, and hyperactivity, are among the most consistently reported behavioral features of Kabuki syndrome across cohorts [10,12,19], and in our patient, they were in fact the presenting concern that prompted psychiatric referral. This matters clinically: hyperactivity in a syndromic, intellectually disabled child is easily read as generic behavioral disturbance rather than a specific, treatable ADHD phenotype warranting standard first-line management. Yet the evidence base for stimulant response in Kabuki syndrome is limited to anecdotal observation, and our patient's paradoxical worsening on methylphenidate, discussed further below, illustrates why treatment expectations should be set cautiously and response monitored closely from the outset.

ASD, anxiety, emotional dysregulation, and self-injury are similarly increasingly reported in Kabuki syndrome and substantially affect adaptive functioning [9,10,12,19], with anxiety appearing to be a core feature rather than a secondary reaction to disability, based on the largest molecularly confirmed behavioral cohort to date [10]. Our patient's presentation, ADHD, ASD, temper tantrums, stereotypies, and self-injury, fits this broader spectrum. Because these symptoms are frequently under-recognized or attributed to intellectual disability alone, we suggest that structured psychiatric assessment, not general developmental follow-up, should be routine in Kabuki syndrome, ideally incorporating standardized autism and behavioral instruments that were not available to us.

Therapeutic reasoning

Methylphenidate was chosen first, consistent with its status as first-line pharmacotherapy for ADHD symptoms even in syndromic intellectual disability. It was discontinued after worsening hyperactivity; paradoxical activation with stimulants is well recognized in children with intellectual disability and ASD and does not, by itself, indicate a syndrome-specific effect.

Aripiprazole was subsequently selected over risperidone, the other antipsychotic with FDA approval for irritability in ASD, to limit sedation, weight gain, and hyperprolactinemia, considerations that carry particular weight in a child already affected by growth restriction and hypotonia. The resulting improvement in agitation and self-injury, despite emergent tremor, is consistent with this drug's recognized efficacy in severe behavioral dysregulation.

Continuing aripiprazole despite tremor, at reduced dose, was considered to be a pragmatic risk-benefit judgment: the tremor was not dangerous or treatment-limiting in itself, whereas the behavioral gain, particularly reduced self-injury, addressed the most functionally impairing and safety-relevant symptom. This decision also had to account for concomitant sodium valproate and clobazam, both of which can independently cause or potentiate tremor, complicating attribution and increasing the importance of close neurological monitoring.

Beyond pharmacotherapy, management combined speech and language therapy and psychomotor rehabilitation, targeting functional communication and motor coordination rather than the underlying syndrome itself. Given the burden of self-injury and tantrums on daily family life, parent-mediated strategies, coaching the mother, who remained the primary informant and caregiver throughout follow-up, in behavioral de-escalation and structured routines, represent a logical extension of this approach, although the specific psychoeducational content offered was not systematically documented in this case. Realistic therapeutic goals in such multisystem, syndromic presentations are rarely curative; they instead prioritize safety, particularly reducing self-injury, functional communication, and caregiver support, with periodic multidisciplinary reassessment as the clinical and cognitive phenotype evolves through adolescence.

Strengths

This report combines detailed DSM-5 psychiatric characterization with systematic dysmorphological, neurological, and genetic evaluation, a combination infrequently reported together in Kabuki syndrome. It also supports current guidance that molecular confirmation, while desirable, is not mandatory for diagnosis, and the *YWHAG* finding contributes a concrete, testable hypothesis, rather than a purely descriptive observation, for future genotype–phenotype research.

Limitations

The Kabuki diagnosis remains clinical, without molecular or epigenetic confirmation. The *YWHAG* variant lacks segregation and functional data. Psychiatric diagnoses relied on DSM-5 clinical assessment without standardized instruments, and treatment response was assessed in routine practice rather than with standardized outcome measures. As a single case, this report cannot establish causality or genotype–phenotype correlation, though it may generate hypotheses for future study.

CONCLUSION

This case reaffirms that a well-characterized clinical gestalt remains a valid and often necessary basis for diagnosing Kabuki syndrome, particularly when systematic exclusion of clinical mimics such as CHARGE syndrome supports it. A negative *KMT2D*/*KDM6A* result on whole-exome sequencing does not resolve every case, and reanalysis, whole-genome sequencing, copy-number analysis, and methylation profiling should be considered before a genetic explanation is abandoned. The coexisting *YWHAG* variant of uncertain significance cannot presently be assigned a causal role, but the documented precedent of blended phenotypes in Kabuki syndrome means it warrants targeted follow-up, including parental segregation testing, rather than dismissal.

Our observation also highlights the importance of systematic psychiatric assessment in children with Kabuki syndrome, as behavioral and neurodevelopmental disorders may represent the principal source of long-term disability and their early recognition can meaningfully guide individualized intervention. This case illustrates how comprehensive clinical phenotyping remains indispensable in the genomic era and suggests that additional rare neurodevelopmental variants deserve consideration as potential contributors to phenotypic variability in clinically diagnosed Kabuki syndrome, pending functional and segregation studies, warranting further collaborative genotype–phenotype research.

Declarations

Ethics approval and consent to participate

Not applicable (single retrospective case report). Written informed consent for publication was obtained from the patient's legal guardian.

Conflict of interest: The authors declare no conflict of interest.

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Data availability

The data supporting the findings of this case report are included within the article. Further de-identified information is available from the corresponding author upon reasonable request.

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