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From neonatal signs to developmental delay: An infant with Wolf-Hirschhorn syndrome – a case study

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Abstract. Wolf-Hirschhorn syndrome (WHS) is a rare genetic disorder with significant developmental, metabolic and craniofacial manifestations, highlighting the importance of continued research due to these challenges. This case report presents an 8-month-old female infant, born at term but classified as small for gestational age, who exhibited developmental delays, metabolic abnormalities and craniofacial dysmorphology characteristic of WHS. This report aimed to emphasise the clinical progression and diagnostic difficulties encountered in cases of WHS. Comprehensive genetic analyses and clinical evaluations were conducted, confirming the diagnosis. Key findings included recurrent respiratory distress with multiple failed weaning attempts, severe failure to thrive, seizures, sensorineural hearing loss, central hypotonia and feeding difficulties. Early neonatal complications and a prolonged, challenging stay in the neonatal intensive care unit were also noted, following which the infant was lost to follow-up until re-presenting at 8 months of age. This report underscores the necessity of early genetic screening and continuous follow-up for infants with congenital anomalies such as WHS. The findings may support paediatricians and neonatologists in the early identification and management of similar cases, thereby improving long-term outcomes

Keywords: 4p16.3 deletion; microcephaly; hypotonia; seizures; failure to thrive; Greek warrior helmet syndrome

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Introduction

Wolf-Hirschhorn Syndrome (WHS) is a rare congenital disorder that presents significant challenges in paediatric healthcare due to its diverse clinical manifestations and the complexity of diagnosis and management. The condition is caused by a deletion of genetic material at the 4p16.3 region and is associated with a spectrum of systemic complications, including neurodevelopmental delays, epilepsy, craniofacial anomalies, cardiac defects and metabolic disturbances. Despite advancements in medical genetics and diagnostic methodologies, WHS remains under-recognised in neonatal and early paediatric care, leading to delays in intervention and the formulation of comprehensive care plans. Given its substantial impact on affected individuals and their families, early genetic screening, multidisciplinary management, and ongoing research are critical to improving clinical outcomes and supporting the development of individualised care strategies.

Seizures are a predominant feature of WHS, often manifesting in early infancy. J. Paprocka *et al.* [1] highlighted that epilepsy in WHS is frequently refractory to conventional antiepileptic drugs, necessitating personalised therapeutic approaches. T. Corrêa *et al.* [2] analysed epileptogenic mechanisms in WHS and demonstrated that multiple genes within the 4p16.3 region contribute to seizure susceptibility. Their findings emphasise the importance of genetic testing in the prediction and management of epilepsy in patients with WHS. Seizures in WHS tend to follow a complex course, often requiring a combination of antiepileptic medications to achieve partial control.

Phenotypic variability in WHS remains a major challenge in clinical diagnosis. E.C. Gavril *et al.* [3] studied seven newly diagnosed cases of WHS, highlighting craniofacial anomalies such as a broad nasal bridge, high forehead and wide-set eyes as the most consistent clinical indicators. A. Mills *et al.* [4] provided insights into the genetic basis of these craniofacial anomalies, demonstrating that neural crest defects play a significant role in the syndrome's characteristic dysmorphology. Their research reinforces the need for early clinical assessment to enable timely diagnosis. Abnormalities in neural crest migration during embryogenesis contribute not only to craniofacial dysmorphisms but also to the congenital heart defects and urogenital anomalies observed in WHS.

Despite advances in medical care, individuals with WHS continue to experience significant morbidity. N.L. Shannon *et al.* [5] examined life expectancy and mortality patterns in WHS, reporting that proactive management strategies are essential for addressing complications such as feeding difficulties, recurrent infections and metabolic disturbances. Feeding difficulties remain a major concern, with many infants requiring nasogastric or gastrostomy tube placement to ensure adequate nutrition. Studies have shown that early nutritional interventions, including specialised feeding strategies, significantly improve growth outcomes and reduce hospitalisations due to failure to thrive.

Cognitive outcomes in WHS vary widely, although intellectual disability is almost universal. A. Battaglia & J.C. Carey [6] emphasised the importance of early intervention using specialised educational programmes and speech therapy, which have been shown to enhance communication skills and overall quality of life. Their study reflects growing interest in targeted educational interventions, including sensory integration techniques and alternative communication methods such as sign language or augmentative communication devices. The psychosocial burden on families caring for individuals with WHS is substantial. S. Berrocoso *et al.* [7] assessed the quality of life and psychosocial features of family caregivers, finding that robust social support networks and effective coping strategies are vital for caregiver well-being. Many families experience considerable stress due to the complex medical needs of children with WHS, necessitating structured caregiver support programmes and access to mental health resources.

This study aimed to document a case of Wolf-Hirschhorn syndrome in an 8-month-old female patient, highlighting the clinical challenges and emphasising the importance of genetic screening and multidisciplinary management in addressing this rare and complex disorder.

Materials and Methods

This case report concerns a patient managed at the Institute of Naval Medicine and Army Hospital (Research & Referral), part of the Hospitals of the Armed Forces Medical Services, India, between February 2021 and June 2023. The patient was monitored and treated over several months, with follow-up assessments conducted during the same period. This report focuses on an 8-month-old female patient diagnosed with Wolf-Hirschhorn syndrome (WHS). Upon admission, the patient underwent a series of clinical evaluations, including a comprehensive physical examination that revealed craniofacial dysmorphisms and developmental delays. Diagnostic investigations included tandem mass spectrometry (TMS) for inborn errors of metabolism, two-dimensional (2D) echocardiography, and brainstem evoked response audiometry (BERA). A high-resolution CT scan of the temporal bones and a magnetic resonance imaging (MRI) of the brain were also performed. To confirm the diagnosis of WHS, clinical exome sequencing was carried out, followed by multiplex ligation-dependent probe amplification (MLPA) to detect a heterozygous deletion of 4p16.3. Both tests were conducted using kits from MRC Holland (Amsterdam, the Netherlands), following the manufacturer's recommendations. The patient was enrolled in early intervention programmes, including physiotherapy, occupational therapy and speech and language therapy. Seizure management involves a combination of antiepileptic drugs (AEDs), including levetiracetam, sodium valproate and clobazam. Nutritional intake was closely monitored, and appropriate measures were implemented to support growth and development. Informed consent was obtained from the patient's legal guardians prior to

all evaluations, treatments and genetic testing. All clinical procedures conformed to the guidelines set out in the Declaration of Helsinki [8]. Written informed consent was also obtained from the patient's parents for the publication of this case report, including the use of clinical photographs. All identifying information has been anonymised to protect the patient's privacy per ethical guidelines and institutional protocols.

Results and Discussion

The case involves a female, the second child of non-consanguineous parents, delivered at full term with a birth weight (Wt) of 2,100 g, length (Lt) of 48 cm, and classified as small for gestational age with asymmetrical intrauterine growth restriction (IUGR) (Ponderal Index: 1.9; weight-for-length: −4.1 Z-score based on WHO chart) [9]. She was initially started on Paladai feeds from day one and remained otherwise asymptomatic alongside her mother until 48 hours post-birth. Subsequently, she developed progressively worsening tachypnoea without significant desaturation. The initial sepsis screen was negative, and chest radiography was unremarkable. Respiratory distress worsened necessitating continuous positive airway pressure (CPAP) management in the neonatal intensive care unit. She experienced a complicated early neonatal course beginning after 48 hours, with difficulty weaning off CPAP. There was no history of rashes, rhinitis, oedema, jaundice, organomegaly, or gross dysmorphisms noted at birth. A detailed antenatal history revealed no febrile illness, bleeding or spotting per vagina, or reduced fetal movements. It was a spontaneous conception; the mother was immunised and had no history of diabetes mellitus or hypertension. TORCH screening was negative, and there was no significant family history. The infant was started on empirical intravenous antibiotics according to the local neonatal intensive care unit protocol, which was discontinued after five days when culture reports returned negative. Multiple attempts at weaning were made, and she was successfully weaned off CPAP after eight days. During this period, she was fed via a nasogastric tube. Following weaning, feeding difficulties emerged in the form of poor oral intake, necessitating a combination of tube feeding and expressed breast milk. This later progressed to partially expressed milk and direct breastfeeding. She experienced weight loss post-CPAP but gradually began to gain weight with direct breastfeeding supplemented by expressed breast milk via Paladai.

Newborn screening with tandem mass spectrometry (TMS) for inborn errors of metabolism was negative. Screening 2D echocardiography revealed a small ostium secundum atrial septal defect, and otoacoustic emissions (OAE) screening showed bilateral "refer" findings at discharge. She was discharged on day 20 with recommendations for follow-up. At discharge, her Wt was 2,200 g (−4 Zscore), Lt was 49 cm (−1 to −2 Z-score), and occipitofrontal circumference (OFC) was 32 cm (−2 to −3 Z-score). She was discharged on direct breastfeeds, and birth-dose vaccines were administered as per the national immunisation schedule. After discharge, she was taken to her maternal home in a rural area in another state and was subsequently lost to follow-up.

Subsequently, the infant experienced recurrent, refractory generalised tonic-clonic seizures beginning at four months of age. A review of the medical records available at that time indicated a weight of 3,600 g (−4.7 Z-score), length of 55.5 cm (at −3 Z-score), and OFC of 36 cm (−3.6 Z-score). She was commenced on antiepileptic drugs (AEDs) – levetiracetam, followed by sodium valproate, which achieved seizure control. At eight months of age, she returned for follow-up with global developmental delay – no neck control, severe failure to thrive, and feeding difficulties (Wt: 4,300 g (−5.2 Z-score), Lt: 60 cm (−3.7 Z-score), OFC: 39.5 cm (−3 Z-score)) – alongside persistent generalised tonic-clonic epilepsy. Seizure semiology revealed that most seizures were triggered by short febrile viral illnesses, typically within the first 24-48 hours of fever onset. Episodes began with a cry, followed by sudden cessation of crying, upward deviation of the eyes, and jerky movements of all four limbs, lasting 2-3 minutes. Minimal frothing at the mouth was noted. Seizure activity either resolved spontaneously or ceased following medication, and was followed by a period of postictal drowsiness. This pattern remained consistent over time.

At the eight-month follow-up, detailed physical examination revealed craniofacial dysmorphism, including a broad nasal bridge extending to the forehead, high anterior hairline with a prominent glabella, highly arched eyebrows, widely spaced eyes, epicanthus, short philtrum, downturned corners of the mouth, microcephaly, and angulated ears (Fig. 1). Haematological and biochemical parameters, as well as metabolic screening, were within normal limits.

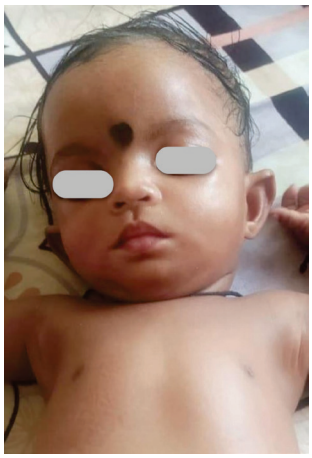

Figure 1. Photograph at eight months of age showing dysmorphic features associated with WHS
Source: authors' photo

She exhibited central hypotonia and bilateral severe-to-profound sensorineural hearing loss, confirmed by brainstem evoked response audiometry, which showed no consistent waveforms (Fig. 2).

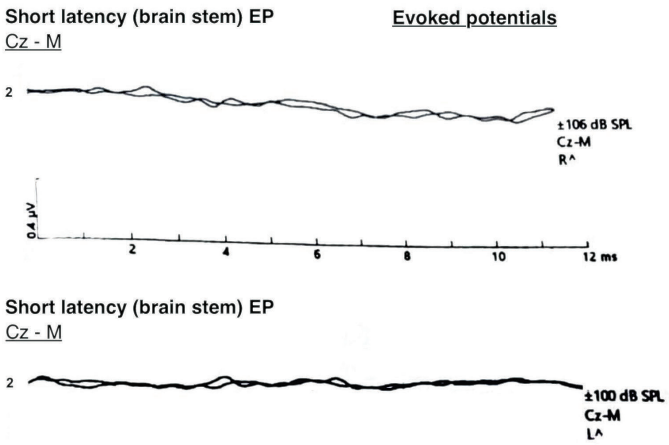

Figure 2. Brainstem evoked response auditory (BERA) showing absence of consistent waveforms
Source: authors' photo

A high-resolution CT scan of the temporal bones revealed no significant abnormalities. In addition, an electroencephalogram (EEG) showed epileptiform discharges in the right parieto-temporal region. Magnetic resonance imaging (MRI) of the brain performed at six months of age showed diffuse thinning of the corpus callosum and a subtle T2 hyperintense signal without diffusion restriction in the inferior aspect of the right cerebellar hemisphere (Fig. 3).

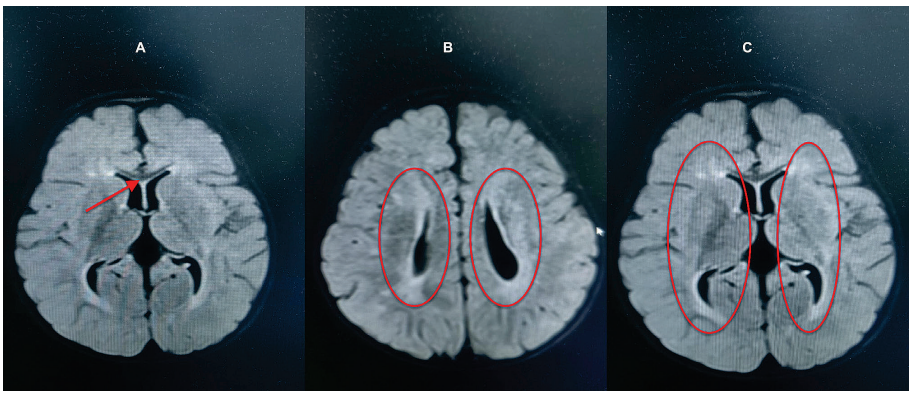

Figure 3. Axial view of brain magnetic resonance imaging
Notes: A – thinned corpus callosum (red arrow); B and C – symmetrical periventricular hyperdensity (red circle)
Source: authors' photo

No significant ocular abnormalities were detected on clinical or ophthalmological examination. The visual evoked potential was normal, indicating preserved cortical response to visual stimuli and intact visual processing pathways. Repeat 2D echocardiography and electrocardiography were conducted to assess cardiac function. Both investigations showed no significant abnormalities, and the previously noted atrial septal defect (ASD) had spontaneously closed, eliminating the need for further cardiac intervention (Fig. 4). A screening ultrasound of the abdomen, kidneys, ureters, and bladder revealed normal findings and chest radiography was also unremarkable.

The patient was lost to follow-up for an extended period, during which her clinical condition underwent significant changes. In view of the dysmorphic features and multisystem involvement, an underlying genetic aetiology was suspected. Consequently, clinical exome sequencing, along with mitochondrial genome sequencing, was performed. Clinical exome sequencing revealed a likely pathogenic copy number variant – a contiguous deletion of 6.2 Mb on Chromosome 4. As the sensitivity and specificity of next-generation sequencing for detecting large deletions and duplications are limited, an alternative method, such as MLPA or chromosomal microarray, was recommended to confirm the deletion. Her craniofacial dysmorphology was suggestive of the Greek warrior helmet appearance. The overall phenotype indicated a possible diagnosis of Wolf-Hirschhorn syndrome. To validate the nextgeneration sequencing findings, MLPA was performed according to the manufacturer's instructions (MRC Holland, Amsterdam, the Netherlands), which confirmed the presence of a heterozygous deletion at 4p16.3 (Fig. 5). The parents were offered karyotyping to rule out a balanced translocation, but they declined further investigation.

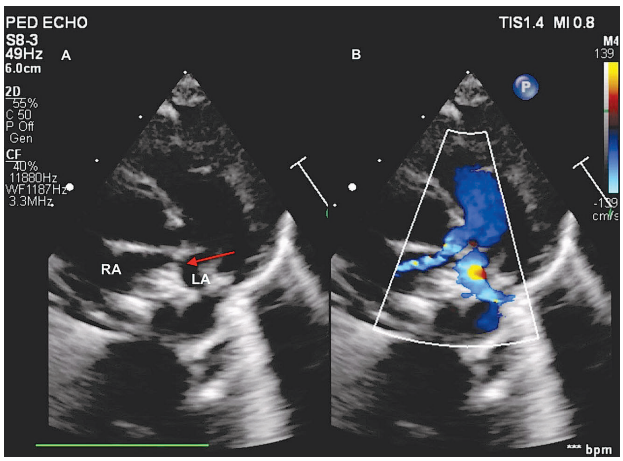

Figure 4. Apical four-chamber view on 2D echocardiography highlighting a small ASD identified during the neonatal period, which resolved spontaneously
Notes: A – ASD depicted by red arrow; B – doppler demonstrating blood flow across the interatrial septum
Source: authors' photo

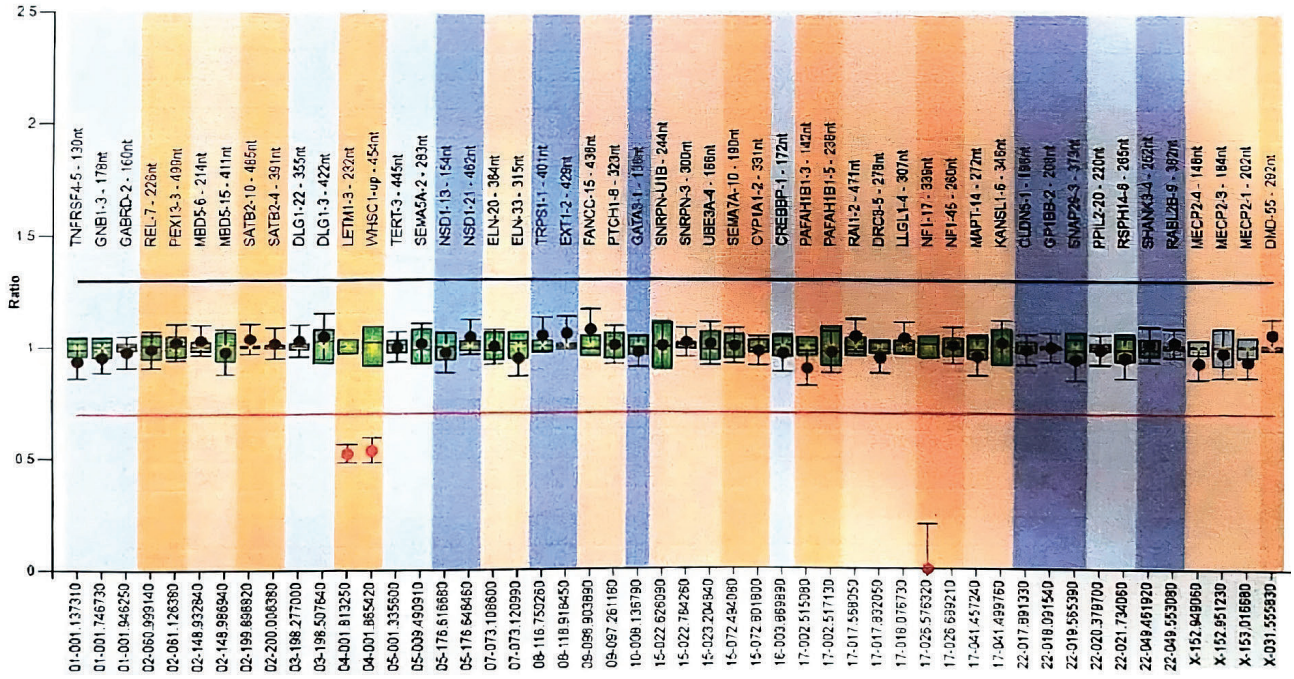

Figure 5. Multiplex ligation-dependent probe amplification (MLPA) analysis
Source: authors' material

She was enrolled in early intervention programmes, including physiotherapy, occupational therapy, and speech and language therapy. She was fully immunised for her age in accordance with the Indian national immunisation schedule. She is currently on three AEDs – levetiracetam, sodium valproate, and clobazam – for seizure control, and continues to exhibit severe global developmental delay. As of July 2024, she is two years and six months old and continues to experience profound developmental delays. Despite intensive rehabilitation, she remains unable to sit independently or achieve other age-appropriate motor milestones. However, her nutritional status has shown notable improvement with tailored dietary interventions, contributing positively to her overall health and growth. Nonetheless, she continues to experience breakthrough seizures, typically triggered by intercurrent illnesses such as viral infections or febrile episodes. These events remain a significant challenge in her management, requiring ongoing adjustments to her antiepileptic regimen. Follow-up EEGs have consistently demonstrated generalised epileptiform discharges, highlighting the persistent seizure activity that complicates her clinical course (Fig. 6). These findings reinforce the need for vigilant monitoring and a flexible, multidisciplinary approach to effectively manage her condition.

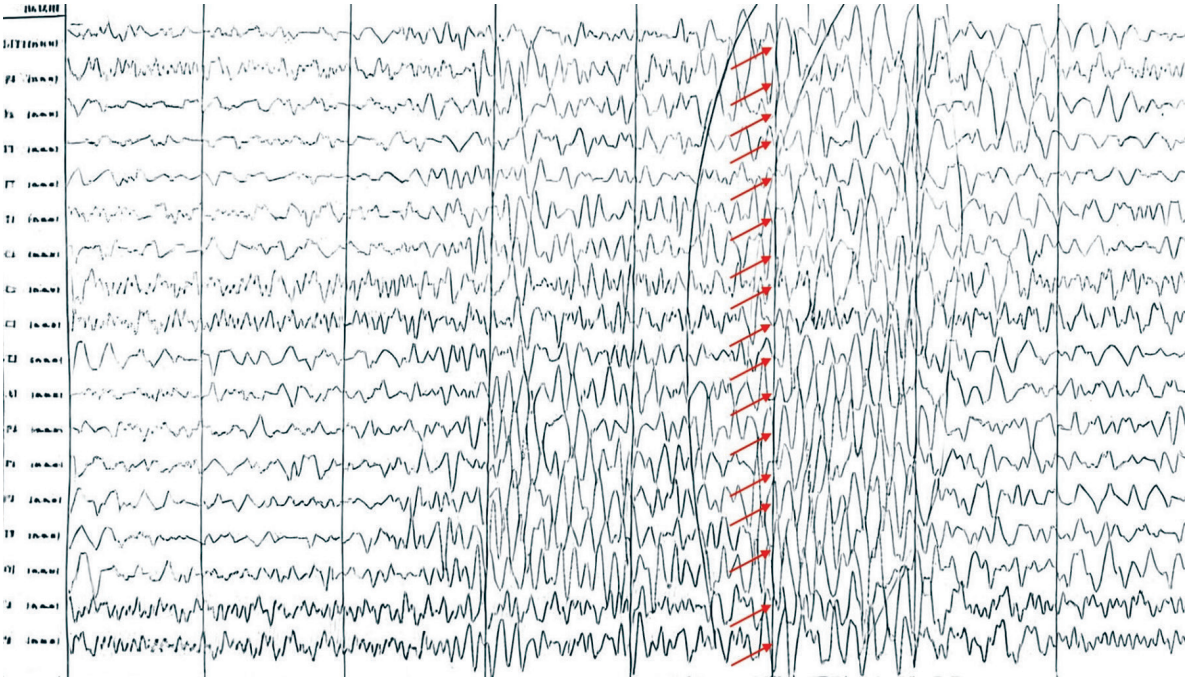

Figure 6. EEG demonstrating generalised epileptiform discharges (red arrow)
Source: authors' material

The management of WHS, as illustrated by this complex presentation, necessitates a multifaceted and highly individualised approach. This case underscores the importance of a comprehensive diagnostic strategy, beginning with the recognition of key phenotypic indicators and extending to advanced genetic investigations. WHS is associated with a wide spectrum of clinical manifestations that vary in severity and presentation depending on the extent of the chromosomal deletion. It is characterised by distinctive craniofacial features, often described as resembling a "Greek warrior helmet" [10]. These features include a broad, flat nasal bridge extending to the forehead, hypertelorism (widely spaced eyes), a high forehead, prominent glabella, and downturned corners of the mouth [11-13]. Other features may include micrognathia, cleft lip, and cleft palate. Such morphological characteristics are essential for the early recognition and diagnosis of WHS.

A study by R. Blanco-Lago *et al.* [14] reported that the mean age of patients in the group was 6.94 ± 6.37 years, while the mean age at diagnosis was 14.34 months. Delayed intrauterine growth was observed in 92.6% of pregnancies. Case management was particularly challenging due to severe developmental delays and the early onset of life-threatening conditions such as refractory epilepsy and profound hearing loss. Epilepsy, a significant concern in WHS, is often well-controlled with appropriate treatment. Generalised tonic-clonic seizures were initially observed, with recurrent episodes occurring despite monotherapy and dual therapy. Eventually, seizure control was achieved with a combination of three AEDs. The seizure type and semiology remained consistent throughout the course of treatment. The selected AEDs – levetiracetam, valproate, and clobazam – were chosen for their efficacy and safety profiles in infants.

A study by A. Battaglia *et al.* [15], involving 87 patients with WHS, revealed that epilepsy occurred in 81 of 87 patients (93%) within the first three years of life. Generalised tonic-clonic seizures accounted for 74%, tonic spasms – 18%, complex partial seizures – 12%, and clonic seizures – 7%. Seizures were often fever-triggered, as observed in many neurological conditions. They frequently occurred in clusters and included status epilepticus in half of the cases. Atypical absences developed in 33% of patients between the ages of one and six years, often with myoclonic components. The EEG showed distinctive abnormalities in 90% of patients, including high-amplitude spike-wave complexes and slow background activity. Epilepsy was well controlled in 81% of cases, primarily with valproate and phenobarbital. Seizure frequency often decreased with age, and many patients became seizure-free. In the present case, seizures were controlled with a combination of levetiracetam, valproate, and clobazam, selected for their established safety and efficacy in infants.

A study by K.S. Ho *et al.* [16], which examined caregiver-reported seizure presentations and treatment outcomes in WHS, highlights several significant findings. Levetiracetam, a broad-spectrum anticonvulsant known to be effective in WHS, demonstrated markedly better outcomes than medications in the carboxamide group. Additionally, both clobazam and levetiracetam showed the highest efficacy and retention rates, making them promising treatment options. These two agents operate via distinct mechanisms of action, providing viable alternatives if a change in therapy becomes necessary. In contrast, carbamazepine,

oxcarbazepine, and phenytoin were associated with the poorest seizure control and the highest incidence of adverse effects [17].

Otolaryngological manifestations of WHS include sensorineural hearing loss, dysplastic ears, deafness, recurrent respiratory tract infections, otitis media, and otitis media with effusion, as described by M.M. Lesperance *et al.* [18]. The patient had profound sensorineural hearing loss; however, due to the severity of the hearing loss and delayed presentation in infancy, it was determined that neither cochlear implantation nor hearing aids would provide meaningful benefit. Although she had no documented evidence of otitis media or related complications, ongoing surveillance during episodes of acute febrile illness is recommended to monitor for such potential issues.

Visual impairments are relatively uncommon in WHS, although some patients experience strabismus, refractive errors, or occasional structural anomalies of the eye. Ocular hypertelorism, ectopia lentis, and iris coloboma are also frequently observed. M.J. Ali & F. Paulsen [19] reported a case of congenital nasolacrimal duct obstruction in WHS, which remains a rare finding. Regular ophthalmological evaluations are essential for the early detection and management of these conditions to support better developmental outcomes. The initial ophthalmological assessment, including visual evoked potentials (VEP), did not reveal any significant abnormalities in this patient. Nevertheless, continued follow-up is necessary to monitor for refractive errors, lenticular abnormalities, and the development of strabismus.

Growth failure in WHS is multifactorial, resulting from feeding difficulties, metabolic abnormalities, and the underlying genetic factors that affect growth. Loss-of-function variants in the WHSC1 gene are associated with developmental delays, autism, and congenital heart conditions, indicating that such variants may lead to a milder phenotype of WHS. Disruptions in WHSC1 contribute to the developmental delays, craniofacial dysmorphisms, and short stature observed in affected individuals. Recent case reports suggest that early nutritional interventions and growth hormone therapy may significantly improve growth parameters in these children. Furthermore, studies by N.J. Boczek *et al.* [20] have demonstrated that consistent monitoring of growth indicators and dietary intake is crucial for timely and effective intervention. Significant growth failure was observed in the present case, primarily due to feeding difficulties that began during the neonatal period and persisted throughout infancy. Recurrent aspiration, often noted in such cases, may result in microaspiration, potentially leading to pneumonia and impaired pulmonary function.

Children with WHS commonly experience nutritional challenges owing to feeding difficulties, which may result in deficiencies of essential vitamins and minerals. A multidisciplinary approach, including input from a dietitian, is essential for devising individualised nutrition plans to address these deficiencies and to promote optimal growth and development. Tailored dietary interventions have been shown to improve weight gain and linear growth in similar cases. A study by T. Antonius *et al.* [21] revealed that all children with WHS exhibited marked intrauterine growth restriction. Despite adequate nutritional support, none demonstrated catch-up growth, and all remained short-statured with profound microcephaly. Given the immunological vulnerabilities often noted in children with WHS, it is essential to maintain an up-to-date vaccination schedule. Specific recommendations include additional vaccines against influenza and pneumococcal infections, as affected individuals may be more susceptible to respiratory complications. Coordination with healthcare providers is vital to ensure compliance with immunisation protocols and to minimise the risk of preventable infections.

A multidisciplinary team – comprising a paediatric neurologist, clinical geneticist, dietitian, and speech and language therapist – was integral to addressing the complex clinical needs of the patient, ranging from seizure control to nutritional support and communication therapy, given the presence of hearing impairment. The paediatric neurologist plays a critical role in diagnosing and managing neurological conditions such as epilepsy**,** which are common in genetic syndromes. The clinical geneticist is responsible for identifying the genetic basis of the condition, clarifying the chromosomal abnormalities, anticipating potential complications, and providing genetic counselling for the family. The dietitian addresses nutritional challenges, often arising from growth retardation, feeding difficulties, or metabolic disturbances, by developing individualised dietary plans to ensure adequate intake. The speech and language therapist focuses on communication difficulties**,** particularly in children with intellectual disabilities or hearing loss. They may introduce alternative communication strategies, including sign language or augmentative and alternative communication devices, to help the child communicate and engage effectively with others. Ongoing research into WHS suggests the potential for targeted therapies to mitigate the effects of gene expression deficits associated with the 4p deletion. Emerging advances in gene therapy and molecular medicine offer hope for more effective treatments that address the underlying genetic mechanisms of WHS. Upon the patient's re-evaluation at eight months of age, a prompt genetic assessment led to the definitive diagnosis of Wolf-Hirschhorn syndrome. Parental counselling was initiated soon after, and a comprehensive multidisciplinary care plan was implemented, incorporating physiotherapy, occupational therapy, speech and language therapy, and an antiepileptic treatment regimen. However, the approach was not without its limitations. The diagnosis was delayed due to a prolonged gap in follow-up following neonatal discharge, and the initial newborn screening was restricted to inborn errors of metabolism, excluding broader genetic testing despite the presence of early clinical warning signs.

Conclusions

This case of Wolf-Hirschhorn syndrome underscores the challenges associated with diagnosing and managing this

rare genetic disorder, which is characterised by multisystem involvement. The findings underscore the importance of early genetic screening, particularly in infants presenting with developmental delays, craniofacial dysmorphisms, or unexplained seizures. The diagnostic process, which included clinical exome sequencing and confirmatory multiplex ligation-dependent probe amplification, identified a 6.2 Mb deletion in the 4p16.3 region. This case demonstrates the utility of advanced genetic testing in confirming complex diagnoses and in guiding clinical management strategies. The management of WHS requires a multidisciplinary approach, given its diverse clinical manifestations, including profound sensorineural hearing loss, severe failure to thrive, and refractory epilepsy. This case illustrates how individualised treatment plans – including the use of antiepileptic drugs such as levetiracetam, valproate, and clobazam – can achieve partial seizure control, despite the inherent complexities of the condition. Early therapeutic interventions, such as physiotherapy and speech and language therapy, proved pivotal in addressing global developmental delays and in improving overall quality of life. Additionally, the case highlights the importance of ongoing follow-up to monitor growth parameters, developmental progress, and the potential for complications such as aspiration pneumonia. The findings emphasise the need for tailored nutritional plans and updated immunisation schedules to reduce the risks associated with immune and metabolic vulnerabilities in patients with WHS. This report contributes to the limited body of literature on this rare condition and provides valuable insights into its clinical course and multidisciplinary management. Future research into targeted therapies and geneediting technologies holds promise for addressing the underlying genetic abnormalities, with the potential to improve long-term outcomes for individuals affected by WHS and related disorders.

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Conflict of Interest
None.

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Від неонатальних ознак до затримки розвитку: немовля з синдромом Вольфа-Гіршхорна – клінічний випадок

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Анотація. Синдром Вольфа-Гіршгорна (WHS) – це рідкісне генетичне захворювання, що супроводжується значними порушеннями розвитку, обміну речовин та черепно-лицевими проявами, що підкреслює важливість продовження досліджень з огляду на ці виклики. У цьому клінічному випадку представлено 8-місячну дівчинку, народжену в строк, але класифіковану як малу для гестаційного віку, яка мала затримку розвитку, порушення обміну речовин та черепно-лицьову дисморфію, характерні для WHS. Цей звіт мав на меті підкреслити клінічний перебіг та діагностичні труднощі, що виникають у випадках WHS. Були проведені комплексні генетичні аналізи та клінічне оцінювання які підтвердили діагноз. Основні ознаки включали рецидивуючу дихальну недостатність з численними невдалими спробами відлучення від грудного вигодовування, затримку росту, судоми, сенсоневральну приглухуватість, центральну гіпотонію та труднощі з годуванням. Також були відзначені ранні неонатальні ускладнення та тривале, складне перебування в відділенні інтенсивної терапії новонароджених, після чого дитина зникла з поля зору лікарів, поки не з'явилася знову у віці 8 місяців. Цей звіт підкреслює необхідність раннього генетичного скринінгу та постійного спостереження за немовлятами з вродженими аномаліями, такими як WHS. Результати можуть допомогти педіатрам та неонатологам у ранній діагностиці та веденні подібних випадків, тим самим покращуючи довгострокові результати

Ключові слова: делеція 4p16.3; мікроцефалія; гіпотонія; судоми; затримка росту; Greek warrior helmet syndrome