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Issue Article

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3D FOOT INVESTIGATION IN CHILDREN WITH ANGELMAN SYNDROME: A CASE-BASED ANALYSIS

Abstract: *Angelman syndrome (AS) is a rare neurogenetic condition often associated with motor impairments and characteristic gait abnormalities. However, detailed assessments of foot structure in affected individuals remain limited. This case-based study presents a comparative analysis of foot morphology in two biological siblings with genetically confirmed AS, using the UPOD Full Foot Scan 3D system. Despite their shared diagnosis, the children exhibited different types and degrees of foot deformities, including asymmetrical arch height, heel valgus, and midfoot instability. The scanning process required methodological adaptation due to behavioral and motor challenges common in AS. The findings underscore the need for early orthopedic assessment in AS and demonstrate the utility of three-dimensional scanning for objective and non-invasive evaluation.*

Key words: *Angelman syndrome, 3D scanning, toe walking, valgus deformity, flatfoot.*

Language: *English*

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Introduction

Angelman syndrome (AS) is a rare neurogenetic disorder resulting from the loss of function of the maternally inherited *UBE3A* gene on chromosome 15q11–q13. It is characterized by severe developmental delays, speech impairment, epilepsy, and a distinctive wide-based, ataxic gait. The estimated prevalence ranges from 1 in 12,000 to 1 in 20,000 individuals.

Although motor deficits and gait abnormalities are frequently observed in individuals with AS, there is a relative scarcity of detailed morphometric data on foot structure in this population. Some reports have described features such as hypotonia, joint hypermobility, pes planus, and orthopedic complications including scoliosis and hip displacement. However, quantitative assessments using modern imaging and scanning technologies remain limited [1-4].

This study presents a rare comparative case of two biological siblings diagnosed with Angelman syndrome. Three-dimensional foot scans were performed using the UPOD Full Foot Scan system (China), allowing for detailed analysis of static foot parameters including arch structure, heel alignment, and forefoot angles.

Importantly, the scanning process in children with AS presents distinct challenges. Cognitive and behavioral characteristics such as hyperactivity, reduced cooperation, and difficulty maintaining still

posture can significantly complicate data acquisition. These factors require careful procedural adaptation and highlight the need for more flexible and child-friendly assessment tools in clinical biomechanics [5].

The objective of this case-based analysis is to document structural foot differences between the two siblings and to contribute to the limited body of literature addressing orthopedic profiles in AS. The findings may support early detection and targeted intervention strategies.

Materials and Methods: This study involved two biological siblings diagnosed with genetically confirmed Angelman syndrome: a 6-year-old girl and her 9-year-old brother. Both children presented with neurodevelopmental impairments consistent with the clinical phenotype of the syndrome.

To assess foot morphology, we used the UPOD Full Foot Scan system (China), a three-dimensional foot scanner designed for static weight-bearing analysis. The device captures high-resolution foot geometry and provides automated measurement of length, width, girth, angular alignment, and arch indices.

Conducting scanning procedures in children with Angelman syndrome presented specific challenges. Due to severe speech and cognitive impairments, participants were unable to understand or follow verbal instructions. Additionally, a common motor behavior in this population—persistent toe-walking—interfered with obtaining standardized weight-bearing positions during scanning. The children also demonstrated limited ability to remain still, leading to motion artifacts or distorted scans in several cases.

To address these issues, multiple scans were performed for each child during a calm state with minimal external stimulation. From the collected data, we selected the most complete and technically reliable scans for the final analysis. Only those scans where the foot was in full plantar contact with the platform and without visible motion artifacts were included. Both left and right feet were analyzed separately for parameters such as arch index, heel angle, hallux angle, malleolar valgus index, and longitudinal arch angle [6-10].

All procedures were conducted under the supervision of a pediatric neurologist and orthopedic specialist to ensure child safety and clinical accuracy.

Results: The analysis revealed significant deviations from normal foot morphology in both children, with clear asymmetry and differing types of structural deformities between the siblings. The key findings are summarized below.

Patient 1 — Female, 6 years old
Right foot:
Arch Index: 0.27 — Low (+)
Heel Angle: 11° — Severe valgus
Hallux Angle: 3.3° — Normal
Malleolar Valgus Index: 10.85 — Planus
Longitudinal Arch Angle: 130.9° — Low
Left foot:
Arch Index: 0.10 — High
Heel Angle: 0° — Neutral
Hallux Angle: 16.0° — Mild valgus
Malleolar Valgus Index: 7.92 — Rectus
Longitudinal Arch Angle: 131.7° — Low

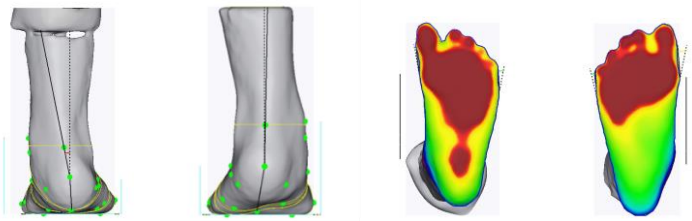

Fig. 1. 3D reconstruction of foot geometry using UPOD Full Foot Scan. Patient 1

Patient 2 — Male, 9 years old
Right foot:
Arch Index: 0.32 — Low (+++)
Heel Angle: 138° — Invalid (likely scanning error)
Hallux Angle: 7.1° — Normal
Malleolar Valgus Index: –50.00 — Invalid
Longitudinal Arch Angle: non-numeric — Invalid
Left foot:
Arch Index: 0.17 — High
Heel Angle: 6° — Moderate valgus
Hallux Angle: 9.5° — Normal
Malleolar Valgus Index: 13.45 — Planus
Longitudinal Arch Angle: 132.3° — Low

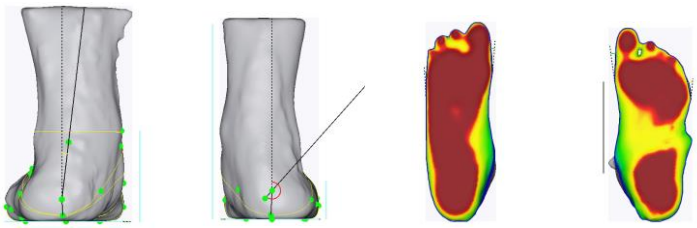

Fig. 2. 3D reconstruction of foot geometry using UPOD Full Foot Scan. Patient 2

General Observations: Both children exhibited asymmetrical foot structure, with valgus deformity and flattened arches more pronounced on the right side. Technical issues, including motion artifacts and atypical toe-walking postures, compromised data integrity for certain right foot parameters, especially in the older sibling. The hallux angles remained within normal limits in both Patients. The variability between feet and between siblings underscores the heterogeneous musculoskeletal presentation in Angelman syndrome.

Discussion: This case-based study presents a comparative analysis of foot structure in two biological siblings with Angelman syndrome (AS), a condition known for its severe neurodevelopmental impairments and motor abnormalities. Using 3D scanning technology, we identified distinct and asymmetric deformities of the feet in both children, highlighting the orthopedic complexity often overlooked in this population.

In the 6-year-old girl, the right foot demonstrated a classic flat-valgus configuration: a low arch index, severe heel valgus (11°), and a “planus” malleolar valgus index. This is consistent with previous clinical observations that children with AS often exhibit pes planovalgus due to hypotonia and joint laxity. In contrast, her left foot had an unusually high arch index (0.10), but a low longitudinal arch angle (131.7°), suggesting an unstable or functionally elevated arch with potential compensatory mechanics. The asymmetry between her two feet is particularly notable and may contribute to altered balance and gait instability.

The 9-year-old boy exhibited more pronounced abnormalities in the right foot, although some values were unreliable due to motion artifacts during scanning (e.g., invalid heel angle and valgus index). His left foot, however, showed a low longitudinal arch (132.3°), moderate heel valgus (6°), and a malleolar valgus index indicative of planus alignment. The discrepancy between the arch index (0.17 = “high”) and the low arch angle again suggests that static footprint-based indices may not always correlate with three-dimensional arch geometry, especially in neurologically impaired children.

Toe-walking behavior, commonly observed in AS and noted in both children during this study, complicates both data acquisition and clinical interpretation. Toe-walking reduces weight bearing on the heel and midfoot, potentially leading to underestimation of rearfoot valgus and misrepresentation of true arch configuration. These postural adaptations may be not only biomechanical but also neurodevelopmental in origin, linked to cerebellar and proprioceptive dysfunction intrinsic to AS.

The clinical implications of these findings are significant. Flatfoot deformity, when combined with heel valgus and midfoot instability, may increase the risk of further functional deterioration, falls, and joint strain over time. Early detection and management—including orthotic support, physical therapy, and serial monitoring—are essential in mitigating long-term disability in children with AS. Furthermore, the observed asymmetry between limbs suggests the need for individualized assessment and intervention, rather than relying on bilateral assumptions.

Despite the small sample size, the study underscores the value of objective morphometric analysis in a population where clinical examination is limited by cooperation and cognitive function. Incorporating 3D scanning into routine care may enhance the accuracy of orthopedic evaluation and support personalized rehabilitation strategies [.

Conclusion: This study highlights the presence of complex and asymmetric foot deformities in two siblings with Angelman syndrome, revealed through three-dimensional scanning. Despite their shared genetic diagnosis, the children demonstrated differing structural profiles, including variations in arch height, heel alignment, and medial-lateral balance. These findings reflect the heterogeneous orthopedic manifestations associated with AS and emphasize the need for individualized assessment.

Toe-walking behavior, poor postural control, and limited cooperation during examination present unique challenges in this population and may lead to underdiagnosis or misinterpretation of musculoskeletal abnormalities. The use of 3D foot scanning offers a noninvasive and objective method to overcome these barriers and obtain reliable data for clinical decision-making.

Early orthopedic evaluation and intervention—including orthotic support and physical therapy—should be considered an integral part of care for children with Angelman syndrome. The implementation of routine foot assessments may help prevent secondary complications, support safer

ambulation, and ultimately improve functional outcomes and quality of life.

Ethical Standards and Protection of Personal Data: All images and data presented in this article were collected and published in full compliance with ethical standards. No identifiable personal information is included, and every effort has been made to protect the privacy and dignity of the children involved.

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