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REVIEW

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Diagnostic challenges of carpal tunnel syndrome in patients with congenital thenar hypoplasia: a comprehensive review

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Abstract

Carpal Tunnel Syndrome (CTS) is the most common entrapment neuropathy, frequently presenting with pain, numbness, and muscle weakness due to median nerve compression. However, diagnosing CTS becomes particularly challenging in patients with Congenital Thenar Hypoplasia (CTH), a rare congenital anomaly characterized by underdeveloped thenar muscles. The overlapping symptoms of CTH and CTS, such as thumb weakness, impaired hand function, and thenar muscle atrophy, can obscure the identification of median nerve compression. This review highlights the diagnostic complexities arising from this overlap and evaluates existing clinical, imaging, and electrophysiological assessment methods. While traditional diagnostic tests, including Phalen's and Tinel's signs, exhibit limited sensitivity in CTH patients, advanced imaging modalities like ultrasonography (US), magnetic resonance imaging (MRI), and diffusion tensor imaging (DTI) provide valuable insights into structural abnormalities. Additionally, emerging technologies such as artificial intelligence (AI) enhance diagnostic precision by automating imaging analysis and identifying subtle nerve alterations. Combining clinical history, functional assessments, and advanced imaging, an interdisciplinary approach is critical to differentiate between CTH-related anomalies and CTS accurately. This comprehensive review underscores the need for tailored diagnostic protocols to improve early detection, personalised management, and outcomes for this unique patient population.

Keywords Carpal tunnel syndrome, Congenital Thenar hypoplasia, Median nerve compression, Diffusion tensor imaging, Nerve conduction studies, Ultrasonography, Magnetic resonance imaging

Introduction

Carpal tunnel syndrome (CTS) is the most common entrapment neuropathy of the upper extremity, caused by compression of the median nerve as it passes through the carpal tunnel [1–4]. Symptoms typically include pain, paresthesias, and weakness in the median nerve distribution, affecting the thumb, index, middle finger, and radial half of the ring finger. CTS impacts a significant proportion of the population, with a prevalence of approximately 1–3% among adults, 3.6 times more common in women than men [1, 5–8]. The condition has occupational and systemic risk factors, such as repetitive wrist

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movements, obesity, diabetes, pregnancy, and rheumatoid arthritis [1, 2].

Diagnosis of CTS relies on clinical history, physical examination, and diagnostic tests [9]. Common clinical signs include the Phalen's test, Tinel's sign, and the Durkan compression test, which have varying sensitivity and specificity [2, 5, 10]. Electrodiagnostic studies and imaging modalities such as ultrasonography and magnetic resonance imaging (MRI) are often used to confirm the diagnosis, especially in atypical presentations or for surgical planning [1, 2, 11–13].

While CTS is widely studied, its diagnosis becomes particularly challenging when associated with congenital anomalies such as congenital thenar hypoplasia (CTH) [14]. CTH is a rare condition characterized by underdevelopment of the thenar muscles. This leads to weakness of thumb opposition, atrophy, and impaired hand function [1, 5]. This anomaly can mask or mimic symptoms of CTS, complicating its diagnosis and delaying appropriate treatment. This review seeks to identify the challenges in diagnosing CTS in patients with CTH and suggest solutions for enhancing diagnostic accuracy.

Methods

A comprehensive literature search was conducted to identify relevant studies addressing the diagnostic challenges of CTS in patients with CTH. The search was performed using the following electronic databases: PubMed, Scopus, and Google Scholar. Keywords and search terms included combinations of "carpal tunnel syndrome," "congenital thenar hypoplasia," "Ultrasound Radiomics," "median nerve compression," and "Diffusion Tensor Imaging." Boolean operators (AND, OR) were employed to refine the search strategy and ensure the inclusion of all relevant articles. Studies published between 1979 and 2025 were included without applying any time restrictions during the search. Inclusion Criteria were Studies focusing on CTS's clinical, imaging, or electrophysiological evaluation in patients with CTH. Case reports, reviews, and original research articles related to the overlap of CTS and congenital hand anomalies.

Articles that did not specifically address the diagnostic aspects of CTS or CTH and were unrelated to the overlapping pathophysiology of CTS and congenital anomalies were excluded. The initial screening process was based on the titles and abstracts, followed by a full-text review of potentially relevant articles. Only English-language articles were included in this review. Additionally, relevant books were also examined as part of the study. Duplicate studies were identified and removed using reference management software. The final selection included studies that contributed significant insights into the clinical presentation, diagnostic methods, and challenges in differentiating CTS from CTH-related symptoms.

Congenital thenar hypoplasia

CTH is classified as a rare congenital hand anomaly that primarily affects the development of the thenar muscles, which are critical for thumb opposition, flexion, and abduction [15, 16]. This condition often occurs as part of radial longitudinal deficiency or as an isolated finding [15]. CTH is typically graded according to Blauth and Buck-Gramcko's classification of thumb hypoplasia [15]. Hypoplastic thumb is classified into five types [15]. Type I represents mild underdevelopment of the thumb, with most functional capabilities preserved and only minor structural or functional changes [15]. Type II involves moderate underdevelopment of the thumb, characterised by significant functional weakness and atrophy of the thenar muscles [15]. Type III includes severe underdevelopment, where thumb function is profoundly impaired, typically requiring surgical intervention [15]. This type is further divided into two subcategories: in Type IIIA, the carpometacarpal joint remains stable, whereas in Type IIIB, instability or absence of the carpometacarpal joint is observed [15]. Type IV signifies the complete lack of the thumb, with no functional capacity [15]. Type V represents the thumb's complete absence and additional hand anomalies, such as radial aplasia, making it the most severe form of hypoplasia [15, 17].

Patients with CTH often present with symptoms that overlap with CTS, such as Weakness in thumb abduction and opposition, Difficulty performing fine motor tasks (e.g., pinching, grasping), and Visible atrophy of the thenar eminence [1, 2, 5, 15, 16].

It can also Alter electrophysiological and imaging interpretations, making isolating median nerve compression from congenital deformities challenging [15]. Delay in diagnosis of CTS, as clinicians may attribute all symptoms to the congenital anomaly without considering concurrent median nerve entrapment [5].

Due to its rarity, the prevalence of CTH is not well established. Still, it is often reported as part of syndromic conditions, including Holt-Oram syndrome, Fanconi anaemia, and another congenital upper limb [Table 1] [1, 2, 15]. Imaging modalities such as ultrasound and MRI are critical in differentiating CTH from CTS-related muscle atrophy. CTH presents with persistent underdevelopment of the thenar muscles, whereas CTS shows progressive changes due to nerve compression [1].

Congenital anatomical variants and nerve compression

Anatomical variants in the carpal tunnel structure or surrounding tissues can contribute significantly to median nerve compression and carpal tunnel syndrome, particularly in patients with CTH. Variants such as anomalous

Table 1 Syndromes associated with hypoplastic thumb

Syndrome	Key Features	Association with Hypoplastic Thumb
Holt-Oram Syndrome [15]	- Autosomal dominant inheritance. - Skeletal abnormalities of upper limbs. - Congenital heart defects.	- Radial ray anomalies, including hypoplastic thumbs. - Often involves thumb underdevelopment or absence.
Fanconi Anemia [15]	- Bone marrow failure. - Physical abnormalities - Increased cancer risk.	- Radial defects, including hypoplastic or absent thumbs. - May present with bilateral thumb hypoplasia.
VACTERL Association [15]	- Non-genetic association of defects: Vertebral, Anal, Cardiac, Tracheoesophageal, Renal, and Limb anomalies.	- Limb abnormalities often involve radial hypoplasia or hypoplastic thumbs. - Thumb anomalies are part of the limb defect spectrum.
TAR Syndrome [15]	- Autosomal recessive inheritance. - Thrombocytopenia (low platelets). - Bilateral absence of the radius.	- Thumb is usually present but may be hypoplastic. - Radial defects can affect thumb development indirectly.

muscles, vascular structures, or fibrous bands may reduce the space within the carpal tunnel, exacerbating pressure on the median nerve [1].

The presence of accessory muscles, such as the palmaris longus, flexor digitorum superficialis, or aberrant fibers of the flexor pollicis longus, can occupy additional space within the carpal tunnel [5, 18, 19]. In some cases, hypertrophy or fibrotic changes in these muscles can further narrow the tunnel, leading to increased median nerve compression [2, 19].

Persistent median arteries or abnormal venous structures within the carpal tunnel may compress the median nerve or contribute to vascular congestion, leading to ischemia [5]. These vascular anomalies are rare but clinically significant, particularly in cases where CTS symptoms are unexplained by other factors [1].

Patients with congenital thenar hypoplasia often have altered ligamentous structures, which can further predispose them to median nerve compression. Fibrous bands or thickening of the transverse carpal ligament can reduce carpal tunnel volume [2, 5].

These anatomical variants play a crucial role in aggravating CTS symptoms in patients with congenital thenar hypoplasia. The combination of limited muscle development, anomalous structures, and nerve compression creates a unique diagnostic and therapeutic challenge [14, 20]. Recognition of these variants through advanced imaging modalities, such as US or MRI, is essential for accurate diagnosis and effective management [1, 19, 21].

Current diagnostic approaches: pitfalls and limitations

Clinical presentation

CTH, a rare condition, is characterised by underdevelopment or absence of the thenar muscles[Figure 1] [22]. This anomaly results in poor thumb opposition, limited pinch strength, and a flattened appearance of the thenar eminence [2].The absence or hypoplasia of the thenar muscles may predispose patients to CTS or mask its diagnosis, as similar motor deficits are observed [5].

Patients with thenar hypoplasia may experience altered biomechanics of the hand, increasing stress on the carpal tunnel and flexor tendons [1]. Diagnostic confusion arises as atrophy, weakness, and functional limitations are often attributed to congenital anomalies rather than concurrent CTS [5]. The pathophysiology of these patients involves intrinsic factors (congenital deficits) and extrinsic factors (nerve compression), requiring a comprehensive understanding of anatomy and detailed clinical evaluation to avoid misdiagnosis [2].

Physical examination

Physical examination remains the cornerstone in diagnosing CTS. Standard clinical tests include Phalen's test, Tinel's sign, and the Durkan compression test. When the median nerve is subjected to compression or stress, these tests elicit symptoms such as pain, numbness, or tingling in the median nerve distribution[Table 2] [23].

Phalen's test The patient maintains wrist flexion for 30 to 60 s. A positive test reproduces CTS symptoms, such as tingling or numbness [24].

Tinel's sign Light tapping over the transverse carpal ligament at the wrist elicits paresthesia in the median nerve distribution [25].

Durkan's compression test Direct compression of the carpal tunnel for 30 s can reproduce symptoms of CTS [25].

While these physical tests have high clinical utility, their sensitivity and specificity are variable. For example, Tinel's sign has a sensitivity ranging from 50 to 75%, and Phalen's test has a sensitivity of 68–73% [23]. In patients with CTH, baseline muscle weakness and altered anatomy may reduce the diagnostic yield of these physical

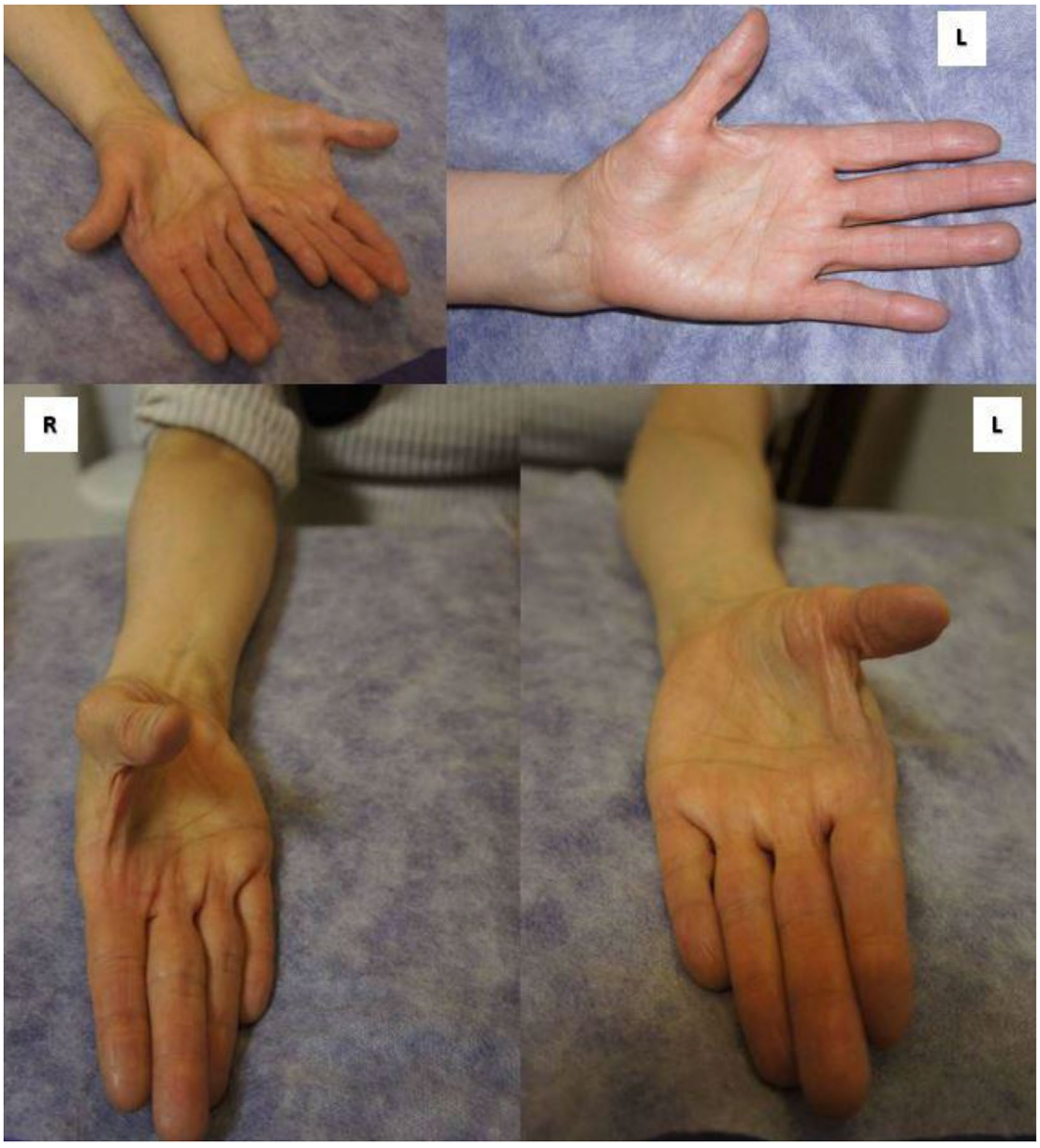

Fig. 1 Clinical presentation of a 46-year-old female referred to our center with a history of hand pain and asymmetric symptoms: right-sided hyposthesia without thenar atrophy, contrasted with left-sided thenar atrophy and limited thumb range of motion but no sensory deficits. The image illustrates the diagnostic complexity of coexisting Carpal Tunnel Syndrome (CTS) and Congenital Thenar Hypoplasia (CTH)

examinations, leading to false negatives or inconsistent results[Table 2] [20, 26].

NCS(Nerve conduction Studies) Nerve conduction studies play a critical role in differentiating CTS from CTH by assessing the latency and amplitude of the median nerve's sensory and motor responses[figure 2] [26]. A prolonged distal motor latency and reduced compound muscle action potential (CMAP) amplitude are hallmark indicators of CTS, reflecting median nerve dysfunction due to compression at the carpal tunnel [26, 27]. However, in CTH, reduced CMAP amplitudes may stem from congenital muscle underdevelopment rather than a neuropathic process, which can complicate diagnosis and lead to misinterpretation of NCS findings [11, 26, 27]. Additionally, sensory nerve action potentials (SNAPs) often remain intact in CTH cases, further distinguishing it from CTS [figure3] [26–28]. To refine diagnostic accuracy, evaluating the first lumbrical muscle, which is also innervated by the median nerve but remains unaffected in CTH, has been proposed as a reliable electrophysiological marker [26, 29]. If CMAP amplitudes are significantly reduced over the thenar muscles but preserved in the first lumbrical, CTH is a more likely diagnosis [26]. Conversely, if both thenar and first lumbrical CMAP amplitudes are

Table 2 Comparison of the effectiveness and limitations of various diagnostic tools for carpal tunnel syndrome in the context of congenital Thenar hypoplasia

Diagnostic Tool	Effectiveness	Limitations in CTH Context
Phalen's Test	Simple, widely used [11, 19, 27] **Sensitivity** 68–73% [11, 19]	Reduced sensitivity due to baseline weakness and altered anatomy [11, 19,27]
Tinel's Sign	Non-invasive, low cost [27] **Sensitivity** 50–75%[11, 19]	Inconsistent responses in congenital cases [11, 19, 27]
Ultrasound	High sensitivity/specificity; identifies anomalies [24, 28, 30] **sensitivity of approximately 95–96%** [24,30]	Confounded by hypoplasia, reduced CSA [28, 30]
MRI	Detailed soft tissue imaging, detects anomalies [26, 29, 32] **Sensitivity** 90–95% [32]	Expensive, less accessible [26, 29, 32]
NCS/EMG	Confirms CTS; evaluates nerve function [26, 32] **Sensitivity** 85–90%[26]	Reduced CMAPs may reflect CTH, not CTS [26, 22]

diminished alongside abnormal SNAPs, concurrent CTS and CTH should be suspected [26].

Recent studies emphasize that incorporating first lumbrical interosseous comparison studies, high-resolution ultrasound, and advanced electrodiagnostic techniques significantly enhances differentiation between these conditions [26, 30]. High-resolution ultrasound (HRUS) can visualize structural abnormalities such as median nerve swelling, compression, or atrophy, providing additional diagnostic clarity when electrophysiologic findings are inconclusive [26, 30]. Furthermore, dynamic nerve conduction techniques, which assess median nerve function across different wrist positions, may provide additional insights into distinguishing CTS-related pathology from congenital anomalies affecting the thenar musculature [26]. These advancements highlight the importance of a multimodal electrophysiological approach, integrating traditional NCS, targeted muscle comparisons, and imaging techniques to ensure accurate diagnosis and avoid unnecessary interventions[Figure 4] [26, 27].

EMG(Electromyography) can assess the electrical activity of the thenar muscles to identify denervation or reinnervation changes [27]. However, baseline abnormalities in CTH, such as absent motor units, can mimic findings seen in advanced CTS, further complicating diagnosis[Table 2] [26, 31].

Proposed solutions and recommendations

Tailored diagnostic protocols

Given the unique challenges in diagnosing CTS in patients with CTH, tailored diagnostic protocols are essential. A combination of advanced imaging techniques, clinical assessments, and careful interpretation of electrophysiological studies can improve diagnostic accuracy.

Advanced imaging combined with clinical history

High-resolution US provides detailed visualisation of the median nerve and surrounding structures. It can identify anatomical variations such as hypoplastic thenar muscles, fibrous bands, or hypertrophied tendons, which are common in patients with CTH [24, 31].

MRI can complement the US by offering high-resolution soft tissue imaging, helping differentiate between congenital hypoplasia and acquired muscle atrophy [32].

Clinical history remains a cornerstone in evaluating symptoms and understanding the patient's baseline deficits. Assessing the progression of sensory and motor impairments over time is critical to distinguishing CTS from static congenital anomalies [26].

Evaluating compensatory mechanisms

Patients with CTH often develop compensatory mechanisms to overcome functional deficits caused by congenital anomalies [15, 20, 33]. Functional assessments, such as assessing grip strength, pinch strength, and manual dexterity, can help identify these adaptations [29].

Combining functional evaluations with imaging findings ensures a more accurate diagnosis and avoids misattributing symptoms solely to congenital anomalies [29, 33].

Interdisciplinary approach

A collaborative, interdisciplinary approach involving multiple specialists is crucial for managing CTS in patients with CTH.

Orthopaedic surgeons can evaluate structural anomalies, such as tendon and ligament abnormalities, and determine if surgical interventions (e.g., carpal tunnel release or tendon transfers) are necessary [1, 6].

Neurologists play a key role in interpreting electrophysiological studies and assessing median nerve function to confirm CTS while accounting for congenital anomalies [26, 27].

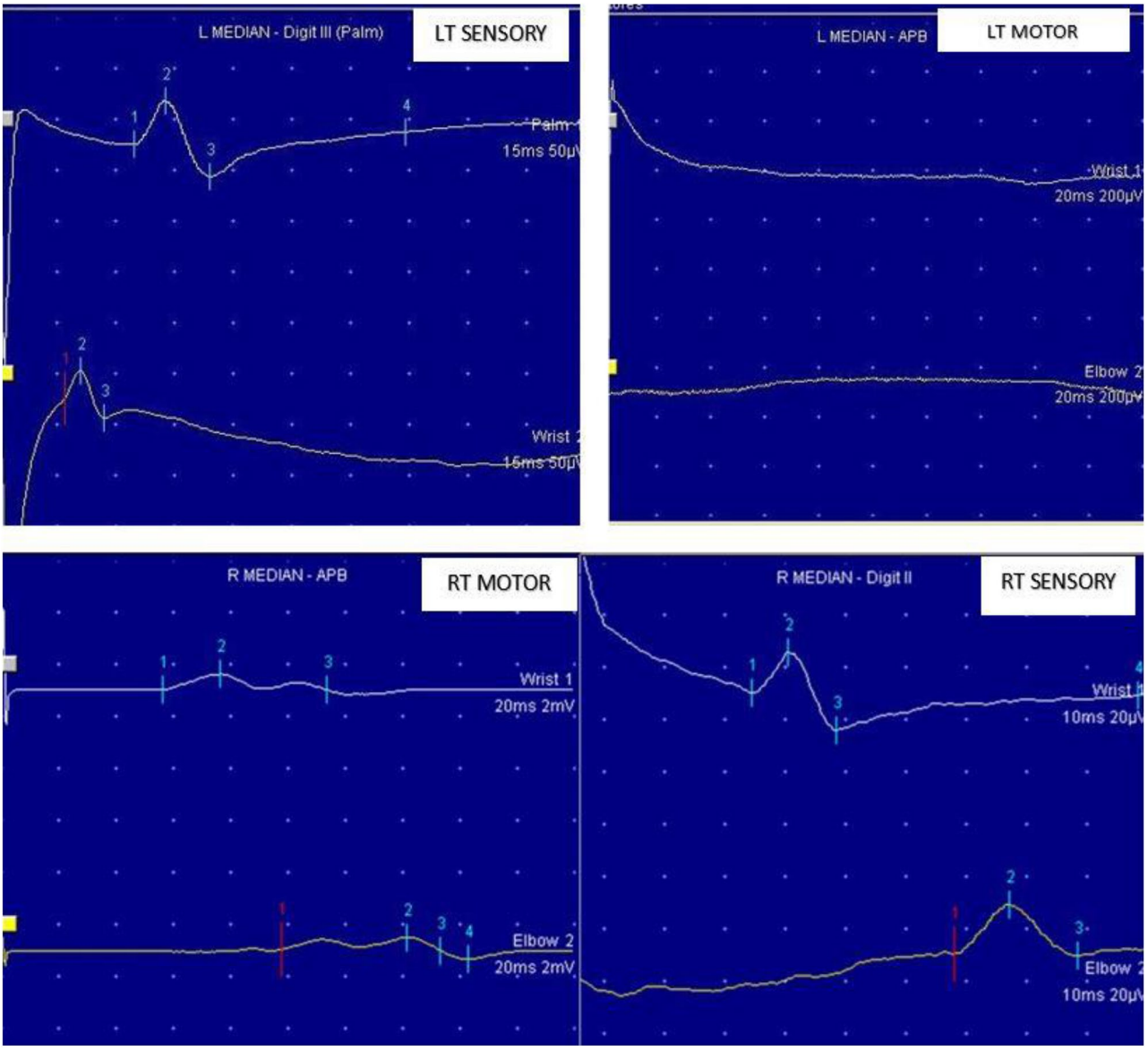

Fig. 2 Nerve conduction study (NCS) and electromyography (EMG) findings for the patient shown in Fig. 1, presenting asymmetric functional impairments. On the left side, sensory nerve responses are intact, confirming normal sensory conduction, but motor responses are completely absent, indicating severe motor dysfunction associated with Congenital Thenar Hypoplasia (CTH). Conversely, the right side shows diminished sensory nerve conduction, consistent with Carpal Tunnel Syndrome (CTS), while motor responses remain intact. These findings highlight the importance of combining clinical, sensory, and motor assessments to distinguish between congenital anomalies and acquired neuropathies in patients with complex presentations

Radiologists contribute by interpreting high-resolution imaging studies (US, MRI) to identify atypical anatomical variations, such as hypoplastic muscles, fibrous bands, or anomalous vascular structures [31, 32].

Physical and occupational therapists can help design rehabilitation programs to optimise function, improve grip strength, and address compensatory mechanisms in patients with CTH [34, 35].

Future directions

Artificial intelligence and machine learning

Artificial intelligence (AI) and machine learning (ML) offer significant advancements in diagnosing and managing CTS, particularly for cases complicated by congenital conditions like CTH [36, 37]. These technologies improve diagnostic accuracy, personalise treatment plans, and reduce errors[Table 3] [36, 37].

AI enhances imaging techniques such as shear wave elastography and quantitative muscle US to identify structural changes in the median nerve and surrounding tissues [36, 37]. Studies show how AI can assist in

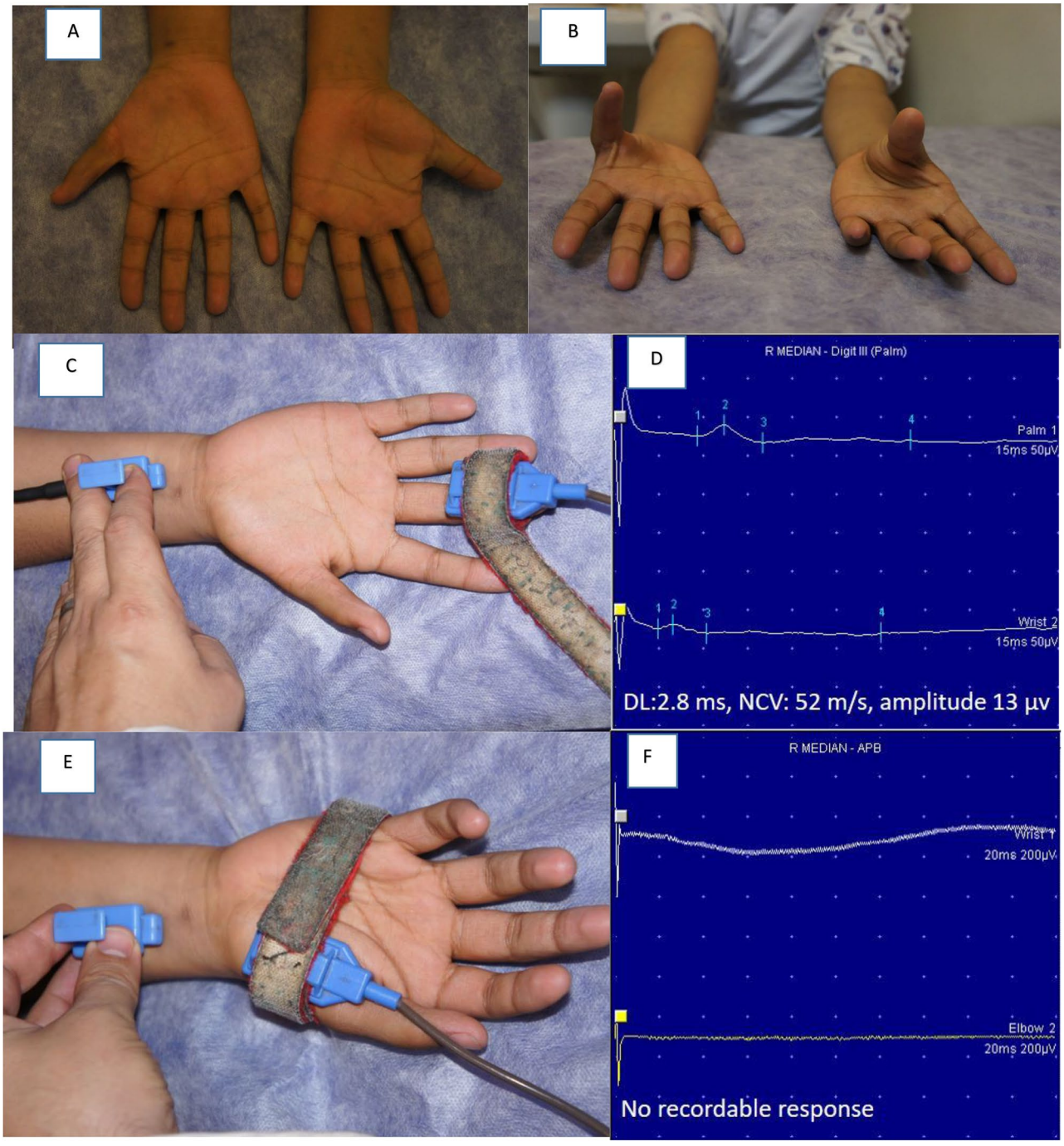

Fig. 3 : (**A**) An 11-year-old patient presenting with thenar atrophy, initially referred with a clinical suspicion of right-hand carpal tunnel syndrome. (**B**) Demonstration of limited thumb motion (**C**,**D**) Sensory nerve conduction study of the median nerve from the third digit showing normal results. (**E**,**F**) Motor nerve conduction study of the median nerve showing **no recordable response**, indicating severe motor dysfunction

detecting early signs of CTS by quantifying muscle stiffness and nerve characteristics, even in complex cases like CTH [36–38].

AI is also helpful in analysing NCS and EMG, automating the process, and improving consistency [27, 36, 37]. This helps differentiate CTS from congenital anomalies by identifying subtle patterns in nerve function [27, 36, 37].

ML models can predict which patients are at higher risk of developing severe CTS [36, 37]. By analysing clinical data and imaging results, AI can stratify patients for more personalised interventions [36, 37].

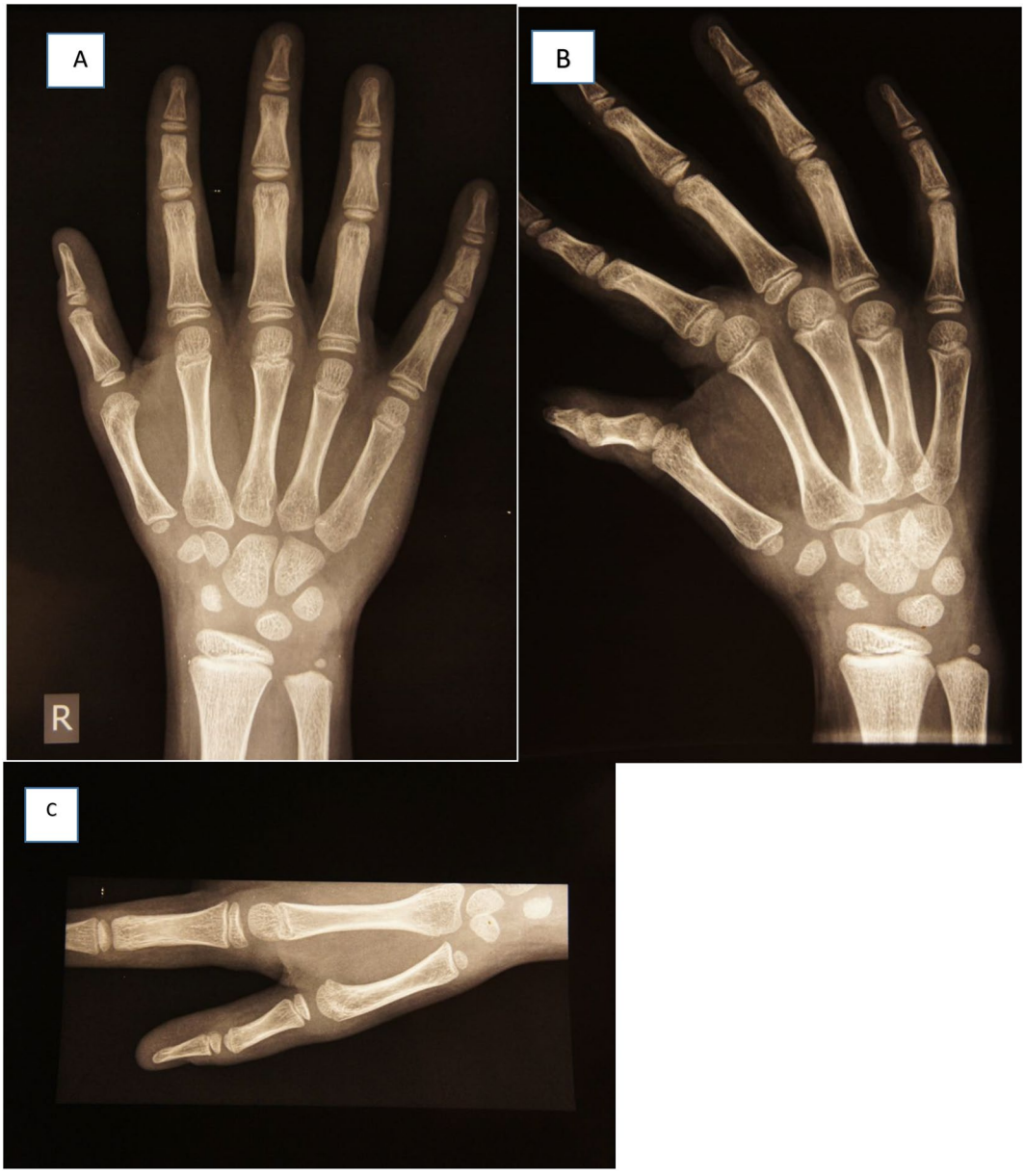

Fig. 4 Radiographic evaluation of the the patient shown in Fig. 3. (**A**) Anteroposterior view, (**B**) oblique view, and (**C**) lateral view demonstrating bony deficiency and varying degrees of instability and subluxation at the first MCP joint. However, the first CMC joint appears stable

AI assists in tailoring treatment plans based on individual patient data [36–38]. Real-time monitoring with AI-powered wearable devices can track changes in nerve compression and muscle activity, enabling early intervention and more precise treatment [36–38].

AI faces challenges such as data quality and model interpretability despite its potential [36, 37]. For AI to be effective in clinical settings, it must be validated across diverse patient populations and provide transparent, actionable insights [36–38].

While AI has shown promising results in diagnosing CTS and grading its severity, several limitations remain. First, AI models require large, diverse training datasets to minimise bias and improve generalizability. Yet, the available data for CTS, especially in rare conditions like congenital thenar hypoplasia, remains limited [36–38]. Second, variability in imaging techniques and electrodiagnostic criteria across studies introduces heterogeneity, making it challenging to establish standardised AI models [36–38]. Third, the integration of AI into clinical practice is hindered by physician acceptance, regulatory challenges, and the need for real-time validation in different healthcare settings [36–38]. Additionally, AI models depend on high-quality input data, and inconsistencies in ultrasound imaging protocols or electromyographic assessments may impact model performance

Table 3 Features, advantages, and limitations of advanced imaging techniques for diagnosing carpal tunnel syndrome and congenital Thenar hypoplasia

Imaging Technique	Description	Advantages	Limitations
Magnetic Resonance Imaging [39, 40, 41]	*Detects subtle changes in the median nerve and surrounding structures *visualizes nerve swelling and structural damage	*High accuracy in grading CTS severity *superior to ultrasonography in nerve morphology assessment	*High cost * limited accessibility *less effective for early dynamic nerve movement analysis
Ultrasonography and Ultrasound Radiomics [44, 45]	*Offers a non-invasive *cost-effective method for real-time visualization of the median nerve and related structures	*Dynamic evaluation during wrist movement * AI-based radiomics enhances diagnostic accuracy and objectivity	*Operator-dependent *may miss subtle or early-stage structural changes
Diffusion Tensor Imaging [39]	*Analyzes nerve fiber integrity by measuring water molecule diffusion along the nerve	*Detects early-stage nerve damage *provides unique microstructural insights	*High cost *limited availability *requires specialized expertise for interpretation
Combination of Imaging Modalities [39, 40, 41]	Integrates MRI ultrasound, and DTI to leverage the strengths of each technique	*Comprehensive diagnostic approach *increases accuracy in complex cases	*Time-consuming *resource-intensive *requires access to all modalities

[36–38]. Addressing these limitations through multi-centre studies, standardised protocols, and explainable AI approaches will be essential for improving the reliability and adoption of AI-driven diagnostics in CTS.

Improved imaging techniques

MRI MRI has become a first-choice imaging modality for CTS due to its ability to detect subtle changes in the median nerve and surrounding structures [39]. MRI has demonstrated high sensitivity (90–95%) and specificity (85–92%) for diagnosing CTS [40, 41]. MRI excels in visualising nerve swelling and structural damage, particularly useful for grading CTS severity [20, 32]. MRI can assess nerve compression and detect early signs of CTS, which are not visible with other imaging methods [40]. MRI is superior to ultrasonography in assessing nerve morphology and grading CTS severity[Table 3] [41].

MRI is less accessible and expensive, but in CTH patients, it can help differentiate between congenital hypoplasia and nerve atrophy due to chronic CTS [32, 39]. Differentiating between congenital hypoplasia and atrophy secondary to CTS remains challenging, as both present with reduced muscle bulk and altered signal intensity [20, 32, 39].

HRUS and US Radiomics: HRUS offers a noninvasive, cost-effective way to visualise the median nerve in real-time [24, 42]. It allows the detection of nerve swelling, compression, and other structural abnormalities associated with CTS [13]. An enlarged median nerve cross-sectional area (> 9 mm^2) at the wrist crease is a hallmark diagnostic criterion for CTS [19, 31, 43]. Yoshii et al. emphasise the US's role in dynamic evaluation, enabling real-time monitoring during wrist movement to reveal nerve compression [44]. The addition of US radiomics, which uses artificial intelligence to analyse image features, further enhances diagnostic accuracy [44]. Faeghi et al. demonstrated that US radionics could provide automated and objective methods for diagnosing CTS, improving traditional radiologist assessments[Table 3] [45].

Studies have reported that HRUS achieves a sensitivity of approximately 95–96% and a specificity of around 94% in diagnosing CTS, reinforcing its role as a first-line imaging tool for CTS and its congenital variants [41]. Recent studies have further highlighted the crucial role of high-resolution neuromuscular ultrasound in diagnosing CTS and differentiating it from congenital thenar hypoplasia [30]. Iyer (2021) demonstrated that HRUS is particularly valuable in cases where electrodiagnostic studies yield inconclusive results due to absent CMAP and normal SNAP, a frequent scenario in patients with CTH [30]. By directly visualising muscle hypoplasia or absence, HRUS enables accurate differentiation between severe CTS and CTH, reducing misdiagnoses that could lead to unnecessary surgical interventions [30]. Furthermore, advanced multiparametric ultrasound assessments, as discussed in recent literature, have been shown to enhance diagnostic precision by integrating multiple ultrasound parameters beyond simple cross-sectional area measurements [46]. This approach provides a more comprehensive evaluation of median nerve pathology, improving sensitivity and specificity in detecting CTS [46]. The integration of these techniques reinforces the role of HRUS as a first-line imaging tool for both CTS and its congenital variants [46].

Diffusion tensor imaging (DTI) is an advanced imaging technique that measures the integrity of nerve fibres by analysing the diffusion of water molecules along the nerve [39]. DTI has emerged as an advanced imaging modality for detecting microstructural nerve damage in CTS. Fractional Anisotropy (FA), a key DTI parameter, has been shown to significantly decrease in CTS patients due to axonal degeneration and myelin sheath damage [39]. Naik et al. (2023) reported that FA values were significantly lower in CTS patients (0.47 ± 0.09) compared to controls (0.61 ± 0.08, $p < 0.001$) [39]. With a cutoff FA

value of 0.54 within the carpal tunnel, DTI demonstrated a sensitivity of 96.4% and specificity of 86.5% [39]. Additionally, FA measurements in the proximal carpal tunnel (cutoff = 0.52) exhibited a sensitivity of 96.2% and specificity of 94.6%, reinforcing the role of DTI in the early and accurate diagnosis of CTS [39].

Naik et al.. Demonstrated that DTI could detect early-stage nerve damage in CTS by analysing the direction and integrity of nerve fibres [39]. DTI provides valuable insights into the microstructure of the median nerve, potentially identifying nerve damage before structural changes become apparent on traditional imaging methods [Table 3] [39].

Combination of imaging modalities Combining MRI, US, and DTI provides a comprehensive approach to diagnosing CTS. Each technique offers unique strengths. MRI is ideal for detailed structural assessment, the US is excellent for real-time dynamic evaluation, and DTI provides insights into the nerve microstructure [31, 39]. Serhal et al. advocate using multiple imaging modalities to increase diagnostic accuracy and enhance treatment planning for CTS, particularly in complex cases [47].

Treatment approaches for coexisting CTS and CTH

Conservative treatments for Carpal Tunnel Syndrome have been widely explored as alternatives to surgical intervention [3]. Studies have shown that noninvasive therapies such as acupuncture, fascial manipulation, low-level laser therapy, cupping therapy, and splinting can significantly improve symptoms and functional outcomes in CTS patients [3].

When CTS coexists with congenital thenar hypoplasia, treatment must address both nerve compression and muscular deficiency [20, 33, 48]. In severe cases where thumb opposition and grip strength are significantly impaired, standard CTS treatments may not be sufficient, and surgical interventions such as tendon transfer procedures may be required [20, 33]. The extensor indicis proprius to opponents pollicis transfer is one of the recommended techniques for restoring thumb function in CTH patients with severe muscle deficiency [20, 33].

For mild to moderate cases, where thumb opposition and motor function remain relatively intact, physiotherapy and compensatory training can help maintain functionality without surgery [20]. Grip-strengthening exercises, proprioceptive training, and adaptive motor strategies may improve hand performance despite congenital muscle deficiency [33].

Future treatment strategies should focus on tailored, minimally invasive approaches. Ultrasound-guided interventions and neuromuscular stimulation techniques may offer new possibilities for non-surgical management [20, 49, 50].

Additionally, minimally invasive procedures like ultrasound-guided carpal tunnel release and suture-based ligament release have shown promise in CTS treatment and could be adapted for CTH patients, though further research is required to validate their long-term efficacy [49, 50].

Limitations

While this study provides valuable insights into the diagnostic utility of various imaging and electrodiagnostic modalities for CTS and CTH-related CTS, several limitations should be acknowledged. First, the rarity of CTH has resulted in a lack of large-scale studies, with most available data derived from case reports and small case series. This inherently introduces selection bias and limits the generalizability of findings.

Second, the included studies are notably heterogeneous regarding study design, imaging methodologies (MRI vs. HRUS vs. DTI), electrodiagnostic criteria, and patient demographics. These variations can influence reported sensitivity and specificity values, challenging direct comparisons across studies. Additionally, the absence of standardised imaging protocols for assessing CTH further complicates consistency in diagnostic performance.

Third, a structured quantitative data synthesis was not feasible due to the limited availability of meta-analytical studies on CTH. While we have incorporated numerical diagnostic performance data from relevant studies, the lack of large-scale cohort studies with standardised methodologies remains a constraint.

Finally, potential publication bias must be considered. Studies reporting significant findings are more likely to be published, while negative or inconclusive results may be underreported. This may overestimate the diagnostic accuracy of certain imaging and electrodiagnostic techniques.

Future research should focus on larger, multi-centre studies utilising standardised imaging and electrodiagnostic protocols to improve the reliability and applicability of findings. Additionally, meta-analytical studies synthesising available data would further strengthen the understanding of optimal diagnostic strategies for differentiating CTH from CTS.

Conclusion

Accurate differentiation of CTS from CTH is essential to avoid misdiagnosis and unnecessary surgeries. Advancements in imaging technologies have significantly improved the diagnosis of Carpal Tunnel Syndrome, particularly in complex cases involving congenital thenar hypoplasia. MRI, HRUS, and DTI provide detailed insights into nerve and structural abnormalities, enhancing early detection and management. Emerging tools such as AI-powered US radiomics might offer automated

and precise assessments, differentiating CTS from similar conditions. Integrating machine learning with imaging modalities promises to transform CTS diagnosis and treatment, paving the way for personalised care and improved patient outcomes. Future research should focus on refining AI-assisted diagnostics and validating multimodal imaging approaches in larger patient cohorts.

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Author contributions
Aidin Arabzadeh: drafted the work, Hamed Naghizadeh: design of the work, Shahram Akrami: interpretation of data, Omid Salkhori: analysis of data, Seyyed Saeed Khabiri: design of the work.

Data availability
No datasets were generated or analysed during the current study.

Declarations

Competing interests
The authors declare no competing interests.

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