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CASE REPORT

Aberrant muscle syndrome: Hypertrophy of the hand and arm due to aberrant muscles with or without hypertrophy of the muscles

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ABSTRACT **Five patients were reported in our congenital anomaly registry who had six hands in total with muscular hyperplasia, aberrant muscles, ulnar drift of the fingers in the metacarpophalangeal (MP) joints, flexion contractures of the MP joints, and enlargement of the metacarpal spaces. Thirty patients with unilateral involvement of this condition have been reported previously. We reviewed these cases and found that the condition varied in severity and that it was reported using different names. However, this condition seems different from true macrodactyly and multiple camptodactyly, including windblown hand, and seems to be an isolated entity of congenital upper limb anomaly. The authors recommend 'aberrant muscle syndrome' or 'accessory muscle syndrome' as a diagnostic name, because this seems to be the most common pathological finding in this condition.**

Key Words: aberrant muscle, accessory muscle, macrodactyly, muscle hypertrophy, windblown hand

INTRODUCTION

Macrodactyly is a relatively rare congenital hand difference. Temtamy and McKusick (1978) classified congenital enlargement of the digit into two groups: one as an isolated anomaly and the other as a part of a syndrome. The former is further classified into true macrodactyly and pseudomacrodactyly. Pseudomacrodactyly means enlargement of the soft tissue of the finger due to hemangioma and congenital arteriovenous (A-V) malformations. In true macrodactyly, the enlargement is mostly caused by soft tissue, which is tumor-like proliferation of the adipose tissue. In this type, the enlarged digital nerves may appear to be infiltrated with fatty tissue. This condition is similar to median nerve lipoma, lipofibromatous hypertrophy of nerves or intraneural lipofibroma (Ogino 1998).

There is another congenital hyperplasia of the hand with or without hyperplasia of the forearm or the whole arm on the same side. It has been reported that this condition is characterized by unilateral muscular hyperplasia, aberrant muscles or accessory muscles, ulnar drift of the fingers in the metacarpophalangeal (MP) joints, flexion contractures of the MP joints, extension contractures of the wrist and enlargement of the spaces between the metacarpals (Mizuoka *et al*. 1962; Yakumaru *et al*. 1980; So 1992; Lanz *et al*. 1994; Tanabe *et al*. 1997; Pillukat and Lanz 2004). In some cases, all these findings appear in the same hand, while in others, only some are present in various combinations. This condition may be pseudomacrodactyly. It is important to distinguish it from macrodactyly, pseudomacrodactyly and windblown hand, as pathological conditions seem quite different from each other (Pillukat and Lanz 2004; Takka *et al*. 2005). The diagnosis of this condition was reported using different names in various papers. We found 33 reported cases of this condition (Lipscomb 1960; Abe *et al*. 1975; Stark *et al*. 1979; So 1992; De Smet & Fryns 1994). In the Japanese modification of the International Federation of Societies for Surgery of the Hand congenital hand anomaly classification, it is called aberrant muscle syndrome, and is classified as failure of differentiation of parts (Ogino 1997). In the present paper, we report our own five cases of this condition (called here aberrant muscle syndrome) and discuss its clinical features and diagnostic name.

CLINICAL REPORT

We found five patients with aberrant muscle syndrome in our congenital anomaly registry. Four cases were male and one female. The right side was affected in one case, the left in three cases, and both in one case. There was no positive family history, or other associated anomalies of the limb and other systems of the body (Table 1).

Case 1: 3-year-old boy
In this patient the left side was affected. The hand was hypertrophic and the MP joints of the fingers were flexed and ulnarly deviated. The thumb was extended and abducted. The palm was bulky (Fig. 1A). Under X-ray, the left hand was hypertrophic, and the enlargement of the intermetacarpal spaces of the left hand was remarkable (Fig. 1B). The humerus, radius and ulna were hypertrophic compared to the unaffected side (Fig. 1C).

Case 2: 3-year-old boy
This was a similar case with flexion and ulnar deviation of the MP joints on the right hand (Fig. 2A). The hand was hypertrophic and the palmar side was bulky (Fig. 2B). Roentgenogram revealed hypertrophic hand, mild ulnar deviation of the MP joints of the fingers and widening of the intermetacarpal spaces. It was wider in the radial side compared to the ulnar side (Fig. 2C). The hypertrophy of the affected hand and arm was remarkable.

Case 3: 3-year, 8-month-old boy
This was a bilaterally affected patient with abduction and extension deformity of the thumb, flexion and ulnar deviation of the MP joints of other digits. The hands seemed hypertrophic and the radial side was bulky. Deformities of both hands were similar, but more severe on the right side (Fig. 3A,B).

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Table 1 Clinical features of cases in the present series of aberrant muscle syndrome

No. cases	Gender	Age (years)	Affected side	Hyperplasia (hand, arm)	Aberrant muscle	Ulnar deviation	Flexion of MP joint	Widening of intermetacarpal spaces	Thumb ext. and abd.
1	Male	3	Left	Arm+	†	+	+	+	+
2	Male	3	Right	Arm+	+	+	+	+	+
3	Male	3	Right	Forearm+	†	+	+	+	+
			Left	Forearm+	†	–	+	+	+
4	Male	2	Left	Forearm+	+	+	+	+	–
5	Female	16	Left	Arm+	+	+	+	+	+

†Surgery has not been preformed;therefore, aberrant muscle could not be confirmed. Abd., abduction; ext., extension; MP, metacarpophalangeal.

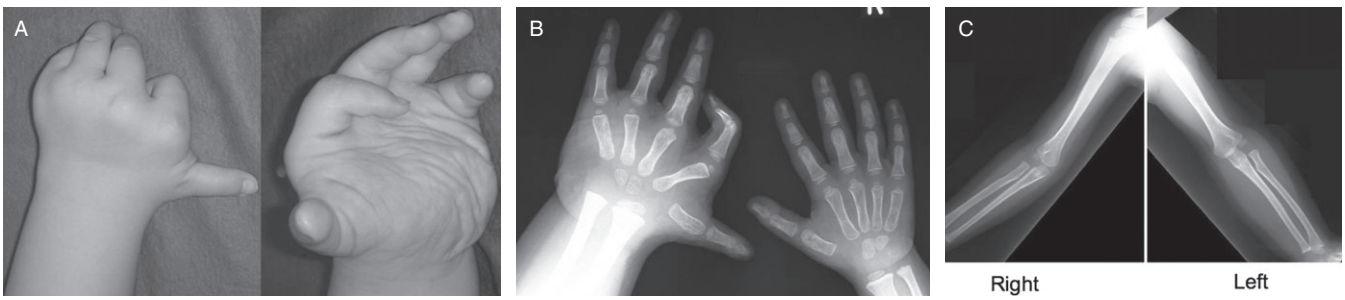

Fig. 1 Case 1: 3-year-old boy. (A) The left hand was affected. The hand was hypertrophic and it looked like windblown hand but there was no thumb in palm deformity. The palm was bulky. (B) Under X-ray, the left hand was hypertrophic, the intermetacarpal spaces were wide and the thumb was extended and abducted. (C) The left arm was hypertrophic compared to an unaffected right side.

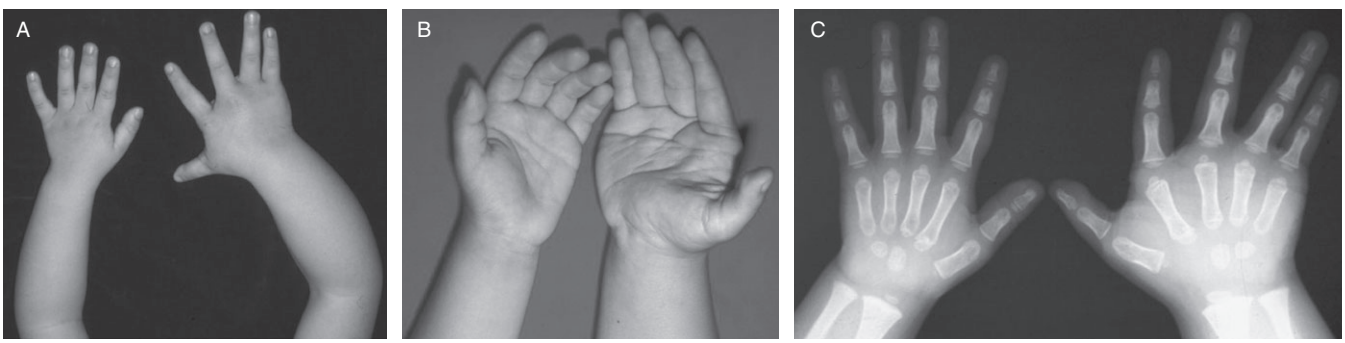

Fig. 2 Case 2: 3-year-old boy. (A) The right hand was affected and the hand and arm were hypertrophic. (B) There was ulnar deviation of the metacarpophalangeal joints of the fingers and the palmar side was bulky. (C) The intermetacarpal spaces were wide and the thumb was extended and abducted.

Case 4: 2-year, 6-month-old boy

This patient had a mild form of the condition. There were flexion deformities of the MP joints of the fingers and a prominence of the hypothenar muscle simulating soft tissue tumor of the hand (Fig. 4A). Roentgenogram revealed hypertrophic hand, ulnar deviation of the MP joints and widening of the intermetacarpal spaces on the right hand (Fig. 4B). Surgery was performed to reduce the volume of the prominence of hypothenar eminence. Intraoperative findings revealed aberrant muscles and hypertrophic intrinsic muscles in the hypothenar eminence. Some were removed but the appearance did not change significantly.

Case 5: 16-year-old girl

In this patient the left side was affected. The deformity of the left hand was treated surgically in another hospital several years before the first visit to our hospital. The operative findings revealed many aberrant muscles. The intrinsic muscles were hypertrophied and disorganized in the hand and forearm. The left

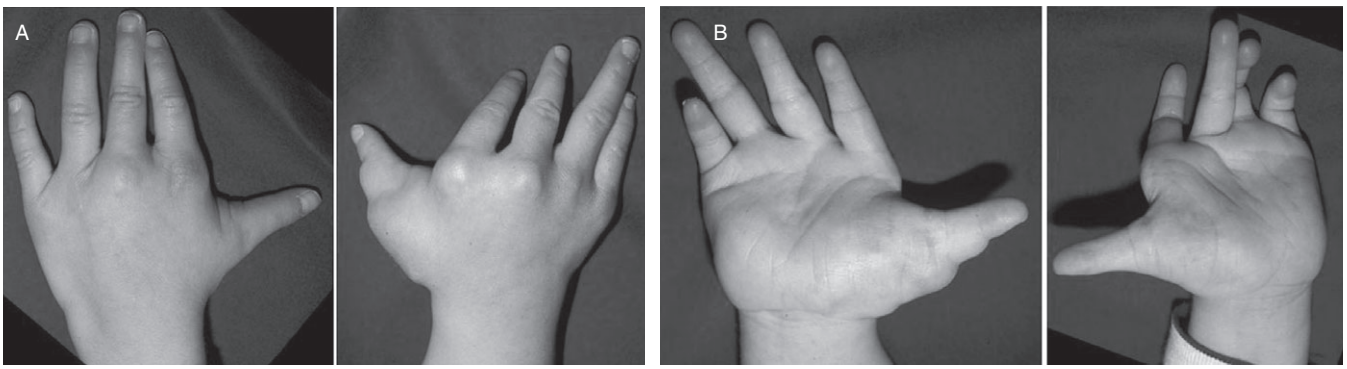

Fig. 3 Case 3: 3-year, 8 month-old boy. (A) Bilateral hands were affected. Dorsal side of the hands: The thumb was extended and there was slight ulnar deviation of the radial fingers at the metacarpophalangeal joints. (B) The palmar side of the hands was bulky in both hands. The thumbs were extended.

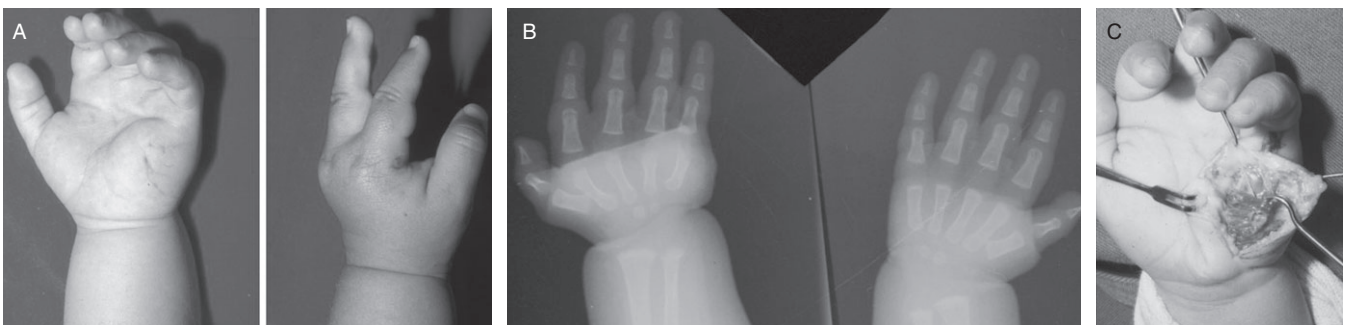

Fig. 4 Case 4: 2-year, 6-month-old boy. (A) The left hand was affected. There was flexion deformity of the metacarpophalangeal joint of the fingers and bulky hypothenar eminence. (B) Roentgenogram revealed hypertrophic hand, ulnar deviation of the radial fingers at the metacarpophalangeal joints and widening of the intermetacarpal spaces on the right hand. (C) Intraoperative finding revealed aberrant muscles and hypertrophic intrinsic muscles in the hypothenar eminence.

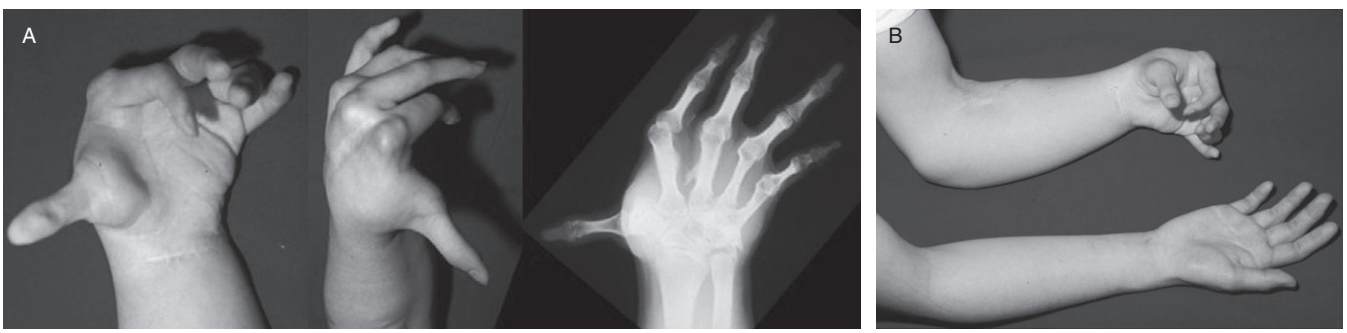

Fig. 5 Case 5: 16-year-old girl. (A) The left hand was affected. The thumb was hyperabducted and the metacarpophalangeal joints of the fingers were flexed and ulnarly deviated. Roentgenogram revealed hypertrophic soft tissues of the hand. The enlargement of the intermetacarpal spaces was not remarkable as metacarpal osteotomies have been performed. (B) The left hand and whole arm were hypertrophic. There was flexion contracture of the elbow.

hand and whole arm were hypertrophic. There was flexion contracture of the elbow and extension contracture of the wrist on the affected side. The thumb was hyperabducted and the MP joints of the fingers were flexed and ulnarly deviated. Roentgenogram revealed hypertrophic soft tissues of the palm with obvious enlargement of the intermetacarpal spaces. This was the most severe case in our series (Fig. 5A,B).

DISCUSSION

The congenital hand difference described in the present paper is characterized by unilateral muscular hyperplasia, aberrant muscles or accessory muscles, ulnar drift of the fingers in the MP joints, flexion contractures of the MP joints, extension contractures of the wrist, enlargement of the spaces between the metacarpals, bulky

Table 2 Clinical features of previously reported cases of aberrant muscle syndrome

Report	No. cases	Gender	Age (years)	Affected side	Hyperplasia	Aberrant muscle	Ulnar deviation	Flexion of MP joint	Widening of intermetacarpal spaces	Thumb ext. and abd.	Other
Lipscomb (1960)	1	M	17	L	+1 (H1)	+1	−1	−1	+1	−1	
Mizuoka *et al.* (1962)	1	M	7	L	+1 (H1)	+1	+1	+	+1	+1	
Abe *et al.* (1975)	4	M:2	6, 10	R:4	+4 (H4)	+4	†(4)	+4	+4	+2. −2	Flexion contracture of the elbow:1
		F:2	11, 15								Crossing of the fingers: 1
Stark *et al.* (1979)	3	M:1	6, 7	R:1	+2 (H2)	+3	†(3)	+1	†(3)	−3	
		F:2	7	L:2	†(1)			†(2)			
Yakumaru *et al.* (1980)	2	F:2	6, 9	R:2	+2 (F1,U1)	+2	+2	+2	+2	+2	Crossing of the fingers: 2
Kayo *et al.* (1982)	1	F:1	13	R:1	+1 (U1)	+1	−1	−1	+1	−1	
Kanie *et al.* (1985)	3	M:2	18, 2	R:3	+3 (F2,U1)	+3	−1	+2, −1	+3	−2	
		F:1	9				†(2)			†(1)	
So (1992)	1	M:1	8	L:1	+1 (U1)	†(1)	+1	+1	+1	+1	
Seto *et al.* (1992)	1	M:1	12	L:1	+1 (U1)	+1	+1	+1	+1	+1	Flexion contracture of the elbow:1
Lanz *et al.* (1994)	3	M:2	10, 12	R:3	+3 (F2,U1)	+3	+3	+2, −1	+3	−3	
		F:1	15								
De Smet & Fryns (1994)	1	F:1	0	R:1	+1 (U1)	†(1)	+1	†(1)	†(1)	†(1)	
Tanabe *et al.* (1997)	1	M:1	2	L:1	+1 (U1)	+1	+1	+1	+1	+1	
Teoh LC	2	M:2	12, 8	L:2	+2 (U1,F1)	+2	+1, −1	+1, −1	+2	+2	
Pillukat & Lanz‡ (2004)	5	M:3	12, 15	†(5)	+5 (H1,F1,U3)	+4	+2, −2	+4, −1	+4, −1	−5	Extension contracture of the wrist: 3
		F:2	19, 18, 15			†(1)	†(1)				Crossing of the fingers: 2
Takka *et al.* (2005)	1	M:1	3	L:1	+1 (U1)	+1	−1	+1	+1	+1	Crossing of the fingers: 1
Present cases	5	M:4	2, 3, 3	R:1, L:3	+6 (F3, U3)	+3	+5, −1	+6	+6	+5, −1	Extension contracture of wrist: 3
		F:1	3, 16	B: 1		†(3)					Crossing of the fingers: 2
Total	35	M:21	0–19	L:13	H:9	+30	+18	+27	+31	+16	
		F:14		R:16	F:10	†(6)	−8	−6	−1	−18	
				B:1	U:16		†(10)	†(3)	†(4)	†(2)	
				†(5)	†(1)						

†( ): number of cases in which the item has not been described; ‡Three of eight cases reported by Pillukat & Lanz (2004) have been already reported by Lanz *et al.* (1994). Abd., abduction; cross, crossing finger when grasping; ext., extension; F, hyperplasia existed in the forearm and hand; H, hyperplasia existed in the hand; L, left; MP, metacarpophalangeal; R, right; U; hyperplasia existed in the whole arm.

palmar surface, and extension and abduction deformity of the thumb (Tanabe *et al*. 1997). While in some patients, the upper arm and shoulder had aberrant and hypertrophic muscles, intrinsic plus position or swan neck deformity and crossing of the index and middle fingers were seen. We found 35 cases, representing 36 hands with this type of deformities in the reported cases and our own series (Tables 1 and 2). Hypertrophy of the hand and/or arm was observed in 35/35 limbs, aberrant muscles in 30/30 hands, ulnar deviation of the fingers in 18/26 hands, flexion of the MP joints in 27/33 hands, widening of metacarpal spaces in 31/32 hands, and thumb extended abducted position in 16/34 hands. The same tendency was observed as in Tanabe's series, although there were variations of the severity. In 1960 Lipscomb reported the mild type as duplication of hypothenar muscle simulating soft tissue tumor of the hand. Case 4 in our series was similar. In the severe type, the whole upper limb was involved (Case 5). Several authors reported that unilateral affection is one of the characteristics of this condition. In fact, all cases reported previously had unilateral involvement. However, Case 3 in our series was a bilaterally involved patient. This suggests unilateral affection is not an essential feature of this condition.

Hypertrophy of the hand is an essential clinical feature of macrodactyly and Proteus syndrome. Hypertrophy of the hand and arm with aberrant muscles has been observed only in aberrant muscle syndrome but not in macrodactyly. The weak point of our paper is that aberrant muscle was only confirmed in the three operated cases. However, this observation was clearly described in all surgically treated cases in the literature (Lanz *et al*. 1994; Tanabe *et al*. 1997; Pillukat & Lanz 2004; Kayo *et al*. 1982; Kanie *et al*. 1985; Seto *et al*. 1992; Teoh *et al*. 2001). Further, there were no other clinical features of Proteus syndrome, such as hamartomatous dysplasia, pigmented nevi and subcutaneous tumors (Lanz *et al*. 1994). Similarities of clinical features among surgically treated and non-surgically treated cases gave us good evidence to suggest that hypertrophy of the hand and/or arm in non-surgically treated cases are also caused by aberrant muscles.

Some of the clinical features of this condition might appear to be in common with other congenital hand differences, including arthrogryposis multiplex congenital, windblown hand, including Freeman-Sheldon syndrome, distal arthrogryposis and macrodactyly (De Smet & Fryns 1994; Wood 1994). Deviation of the MP joints of the fingers is also an essential clinical feature of windblown hand. Wood (1994) reported that the term windblown hand represents a conglomerate of syndromes and different anatomical causes. This concept has led De Smet and Fryns (1994) to propose that aberrant muscle syndrome was congenital windblown hand deformity presenting as the first clinical sign of Proteus syndrome. They thought that the case reported by So (1992) might be the same condition. In windblown hand, thumb in palm deformity is an essential feature and the thumb is always in flexed and adducted position (Ogino *et al*. 1993), but in aberrant muscle syndrome, mostly the thumb is in extended and abducted position as shown in our series. In a case reported by So, the thumb was also in extended and abducted position. Lanz *et al*. (1994) reported ulnar deviation of the fingers in their cases, but they considered it was a different entity from the previously described 'windblown hand'. Grünert *et al*. (2004) also thought that there are two types of windblown hand. One is typical bilateral flexion-adduction contracture of the thumb along with ulnar deviation at the MP joints of the fingers. The other is a unilateral involvement with limb hypertrophy. They also reported that in the latter type, abnormal and hypertrophic muscles were routinely found and it must be distinguished from the typical type. In windblown hand, the palmar skin is always tight, but in this condition it is redundant. In typical windblown hand, there is no widening of the intermetacarpal spaces, which are commonly seen in this condition. This condition seems different from true macrodactyly and windblown hand and does seem to be an own isolated entity of congenital upper limb anomalies. Based on the analysis of the clinical features of this condition, the authors believe that irregularly hyperplastic and aberrant muscles might be the underlying pathologies and may cause hyperplasia of the hand and arm associated with windblown hand deformity.

As for the diagnostic name of this condition, it was reported using different names. In the literature, the first case was described by Lipscomb in 1960. In that case, there was no ulnar deviation of the MP joints of the fingers, but surgical findings revealed that hypertrophic muscles and aberrant muscles caused the enlargement of the hand. The second case was reported by Mizuoka *et al*. in 1962. The same condition was reported as congenital deformity of the arm and palm, with specific features by Abe in 1975 in Japanese. At that time, terminology of this condition was discussed by the Japanese Society for Surgery of the Hand, and aberrant muscle syndrome was proposed as a diagnostic name. Different descriptive terms have been used (Stark *et al*. 1979; Yakumaru *et al*. 1980; So 1992; De Smet & Fryns 1994; Lanz *et al*. 1994; Tanabe *et al*. 1997; Pillukat & Lanz 2004), but we need common terminology. The authors recommend 'aberrant muscle syndrome' or 'accessory muscle syndrome', because these seem to be most common pathological findings in this condition and the terminology is simple.

CONCLUSION

Congenital unilateral muscle hyperplasia of the hand with ulnar deviation of the fingers seems to be an own isolated entity of congenital upper limb anomalies. The underlying pathologies of this deformity might be irregularly hyperplastic and aberrant muscles. 'Aberrant muscle syndrome' or 'accessory muscle syndrome' is recommended as a diagnostic name for this condition.

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