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**Authors:** Krzysztof P. Dąbrowski, Paweł Palczewski, Hanna Stankiewicz-Jóźwicka, Arkadiusz Kowalczyk, Jarosław Wróblewski, Bogdan Cizek

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## **A fully capable pianist with a congenital bilateral agenesis of extensor pollicis brevis muscle**

Krzysztof P. Dąbrowski et al., A fully capable pianist with a congenital bilateral agenesis of EPB muscle

Krzysztof P. Dąbrowski<sup>1</sup>, Paweł Palczewski<sup>2</sup>, Hanna Stankiewicz-Jóźwicka<sup>3</sup>, Arkadiusz Kowalczyk<sup>1</sup>, Jarosław Wróblewski<sup>3</sup>, Bogdan Cizek<sup>1,4</sup>

<sup>1</sup>Department of Descriptive and Clinical Anatomy, Center for Biostructure Research, Medical University of Warsaw, Poland

<sup>2</sup>First Department of Clinical Radiology, Medical University of Warsaw, Poland

<sup>3</sup>Department of Instrumental Studies, The Fryderyk Chopin University of Music, Warsaw, Poland

<sup>4</sup>Department of Neurosurgery in Bogdanowicz Children's Hospital, Warsaw, Poland

Address for correspondence: Krzysztof P. Dąbrowski, Department of Descriptive and Clinical Anatomy, Center for Biostructure Research, Medical University of Warsaw, ul. Indiri Gandhi 35/15, 02-776 Warszawa, Poland, e-mail: [krzysztof.dabrowski@wum.edu.pl](mailto:krzysztof.dabrowski@wum.edu.pl)

### **Abstract**

A 28-year-old male musical student has been presented with visible inability of active abduction and extension of the thumbs in both hands beyond the neutral position. The student has not been previously diagnosed and claimed no history of trauma or surgical procedures in the area of hands and no family history of such disabilities. The student remained capable of playing on keyboard instruments on high level due to compensation by hyperextension of the interphalangeal joint of both thumbs and showed no increased frequency of the injuries or playing-related disorders. The ultrasound and MRI imaging showed complete bilateral agenesis of Extensor Pollicis Brevis muscles and was classified as isolated Congenital Clasped Thumb Syndrome. Due to the age of the student and the agenesis of the muscles the

conservative treatment was deemed inadequate and due to high functionality of the student as a musician and unforeseeable results it might have on a musician's career, surgical treatment has been disadvised.

**Key words: extensor pollicis brevis, hand, musician, agenesis, clasped thumb**

## **INTRODUCTION**

Extensor Pollicis Brevis muscle (EPB) is one of the newest – from an evolutionary point of view – structures in the upper extremity. As a separate, independent muscle it seems to only exist in humans and gorillas [9] which might be one of the reasons behind its substantial morphological variability, so much so it can have significant differences even between the two hands of the same individual [5]. In its standard description EPB has its origin on the posterior surface of the radius and the adjacent part of the interosseous membrane of forearm, dorsally to the origin of Abductor Pollicis Longus muscle (APL). The tendon of the EPB passes through the first compartment of the Extensor Retinaculum and has its insertion at the posterior surface of the base of the proximal phalanx of the thumb [5, 7, 9].

Among most common variations of EPB there are differences in size of the belly of the muscle, including significant reduction, size and shape of the tendon including splitting into multiple collagenous bands, separation of the tendon of EPB from the tendon of APL within the compartment of the Extensor Retinaculum, varying degree of fusion with the APL and changes in exact location of the insertions [4, 6, 7, 9]. Singular authors include also complete duplication of the muscle [4], or complete agenesis of the muscle [9], however those statements do not seem to be reflected in other authors' work, or sometimes even in those same authors studies.

The matter of function of the EPB proves to be a little more complicated than it could be assumed. Although it is described to be an extensor of the first metacarpophalangeal joint, as the name would suggest, the EPB seems to play an important part in the abduction of the thumb, function and structure of the extensor hood of the thumb and extension of the interphalangeal joint of the thumb [5, 10]. And yet, at the same time there are authors arguing that EPB's contribution to the mobility of the thumb is so limited that its reduction or even full absence should not cause significant limitation in function of the hand [6] It depends

however on what can be considered a „significant” limitation, especially when it comes to a profession depending on quick, repetitive and precise movements of the hand, thumb included.

## **CASE REPORT**

During data gathering for a different study a 28 year old male student of musical university has been brought to authors’ attention. The student has been complaining about straining of the thumbs, especially during exercises and repertoire requiring wide spreading of the hand and the teacher of the instrument have noticed unusual shape and function of the student’s hands.

A general examination, a face to face semi-structured interview and an observation of the work with the instrument were arranged. Upon general examination thumbs of both hands showed symmetrical limitation of the extension and abduction in active movement with inability to surpass the neutral position in which the long axis of the proximal phalanx of the thumb is more or less parallel to the forearm , which the student compensated with near 90 degrees overextension in the interphalangeal joints and a very strong ulnar adduction of the hand and abduction of the fifth finger. Examination of passive movement shown a full range of motion without resistance of the tissues. Patient denied any history of substantial trauma or surgical procedures in the area of the hands and wrists and claimed that aforementioned limitation was present for as long as he could remember. Family history showed absence of similar limitations in close family members, although due to personal reasons gathering information on the extended family was impossible.

Interestingly, according to the student, despite going through full musical education from the early years of the childhood, the teacher at the University level was the first one to notice and question the described limitation of the hands’ function.

According to the teacher, the examined student does not show any decrease in ability to play compared to his peers and does not show increased frequency of strain or misuse related trauma.

Following general examination and history the student has been examined via ultrasound (LOGIQ

F8 GE with L6-12, 6-13MHz probe). The examination showed bilaterally a single tendon in the first compartment of the Extensor Retinaculum which was recognized as belonging to APL. Neither a tendon nor a belly of the EPB were found.

For the sake of confirmation an MRI imaging was performed with a 1.5T scanner (Ingenia, Philips, Eindhoven, the Netherlands) using a body coil and a dedicated 8-channel phased-array wrist coil (ds Wrist). Three-millimeter thick T1-weighted FSE images without fat suppression of both forearms and wrists were acquired in coronal and axial planes (Table 1).

MRI showed bilaterally sole tendons of APL in the first compartments of Extensor Retinaculum. The muscle bellies of the EPBs were absent, their anatomic locations were filled by the APL bellies. The remaining forearm muscles were unremarkable.

## **DISCUSSION**

The matter of morphological variability is one of the key aspects of anatomical studies, and not without a reason. Throughout the world medical practitioners learn human anatomy based on textbooks and atlases showing standardized, idealized versions of each structure and each organ and only some of them describe a few most common variations, which is often considered an extracurricular material. At the same time research shows that a substantial proportion, in not majority, of clinical malpractice claims within surgical specialties is a direct result of operator's ignorance regarding morphological variability [6]. It is safe to assume that the situation is similar when it comes to conservative treatment as well.

The clinical image observed in the examined student seems to fall within a category of a „congenital clasped thumb syndrome” (CCTS) which is defined as a congenital flexed and adducted thumb caused by deformity or agenesis of the muscles of the thumb, usually the extensors. In most cases it is diagnosed after the third month of life, due to the natural position of the thumb within the hand but it can also be diagnosed via radiological imaging or the autopsy. [8, 11, 13]. It is usually observed as an accompanying syndrome in serious genetic dysfunctions. Subasioglu observed that in 88% of cases CCTS is associated with serious deformities related to genetic disorders and mentions 27 genetic disorders that consistently feature CCTS [11], for example Tae describes a case of CCTS in a newborn with sirenomelia [12].

Weckesser theorizes that to maintain such frequent symmetry CCTS would have to be associated with a genetic defect within the zygote before first division and suggests its origin in a recessive, sex-linked gene since, according to his observation supported later by Ghani, majority of cases are observed in males [2, 14].

In cases when CCTS is an isolated deformity, or patient's condition is good enough for CCTS to be considered a priority there are treatment options to be considered. Most common and most effective treatment is a surgical tendon transfer of either EPB if possible, or Extensor Indicis muscle. It is considered to have satisfactory results allowing for regaining full range of motion and ability to perform everyday tasks, and small chance of post-surgery complications. There is also an option of conservative treatment of splinting followed by manual therapy. Unfortunately, it is considered applicable only in very mild cases and with the youngest of patients – Al Smail in his case report described a five year old girl as too old for conservative treatment – it is also considered ineffective in cases of complete agenesis of EPB [1, 8].

Regarding the case described in this paper it needs to be said that it is a very unique occurrence, beginning with full bilateral agenesis of EPB without any confirmed family history. Ghani and Soubhagya in their writing acknowledge possibility of complete agenesis of EPB but also state that they have not observed such occurrence in their studies. Rousset describes only one case when EPB was replaced by a ligamentous band from the radial styloid process to the proximal phalanx of the thumb [2, 7, 9]. Most described cases of CCTS have confirmed family tendency [8, 14] yet that is not the case. Another matter is lack of any other visible anomalies. As it was stated before, CCTS is usually considered a symptom accompanying other genetic dysfunctions. According to Ghani in 78% of cases of CCTS other dysfunctions are clearly noticeable during general examination [2] and considering existing research it would not be unreasonable to expect the isolated CCTS cases to be related to reduction or weakening of EPB rather than full agenesis. That being said, Al Smail describes a case of a 5-year-old girl with full agenesis of both extensors of the right thumb with full stability of the carpometacarpal joint of the thumb and lack of any other malformations or anomalies [1]. Hong on the other hand describes a case of a 10-year-old girl with full agenesis of Abductor Pollicis Longus, Abductor Pollicis Brevis, Opponens Pollicis and Extensor Pollicis Brevis in her right hand and a surprisingly efficient compensation by Extensor Pollicis Longus thanks to unusual placement of the insertion on the distal phalanx which allowed the abduction and extension movements to reach the neutral position. Hong describes

function the thumb as „satisfactory” [4]. In this report however, there is a 28-year-old keyboard musician that despite visible limitations in movement is capable of performing at consistently high level and up to that point remained undiagnosed due to efficient compensation mechanisms which, arguably, qualifies for a much better mark than „satisfactory”.

Another surprising factor was the student being undiagnosed throughout the primary and secondary musical education despite being tutored by multiple teachers of multiple instrument, including piano, trumpet and church organs which might suggest that many teachers on the primary and secondary level do not have the basic anatomical knowledge that could be helpful in taking care for their students health, nor do they give enough attention to their technique and biomechanics of their hands as long as the score is being played adequately. It was only at the level of higher musical education that a teacher has noticed irregularities. It could be due to a very effective compensation mechanism of the student, including near 90 degrees overextension in the interphalangeal joints and a very strong ulnar adduction of the hand and abduction of the fifth finger resulting in a hand-span of 21cm in the left hand and 19.5cm in the right between the tips of the first and fifth finger in maximal active stretch. Although it is a modest span compared to the size of the student’s hands, it does not stand out much among his peers. Thanks to that the student has been able to consistently perform on a high level.

A question can be posed „what can modern medicine do in that case”. Considering that the examined student’s thumbs were capable of a full range of movement when moved passively, it would fall under type 1 both in Weckesser’s and Tsuyuguchi’s classification [13, 14] which can be considered a mild case of CCTS. Yet both the age of the student and the fact of complete agenesis of EPB disqualify the idea of treatment through splinting and therapy, especially since such proceedings would ban the student from playing for a prolonged period of time with little to no reward in sight [1, 8]. It is not possible to strengthen an absent muscle. An intervention via orthosis or therapy on the present muscles were discussed and the authors decided against it, seeing as it would carry a risk of disrupting the compensation mechanism. The authors of this paper would also like to make an argument against surgical treatment due to foreseeable risks. Although in most cases the surgery is safe and brings good results, it is important to consider that most patients undergoing such procedure are children and the criteria for a successful operation fall within the spectrum of ability to write and perform everyday tasks, not performing piano recitals. The authors find no reasons to believe that an

invasive procedure requiring long convalescence and therapy by design could improve a quality of life and career longevity of the discussed musician.

Nevertheless, there are certain risks that need to be taken under consideration regarding the student's further career. The full scope of occupational risks and diseases of musicians is not yet known and increase of these risks in case of such an unusual case is hard to estimate. Henry describes a tendency for osteoarthritis to affect the carpometacarpal joint of thumb, causing adduction contracture of the thumb and – when forced against in everyday use - resulting in hyperextension instability and significant deficits in function [3]. It is something to be wary of in future management of the examined student since described mechanism shares many similarities with the student's compensation mechanism.

Considering the brought up arguments it seems reasonable to suggest a close observation and care of a trusted medical professional to facilitate early treatment and management of any disorder that might occur in the future career and continuous work with an instrument teacher in order to choose a repertoire that causes the least possible amount of strain.

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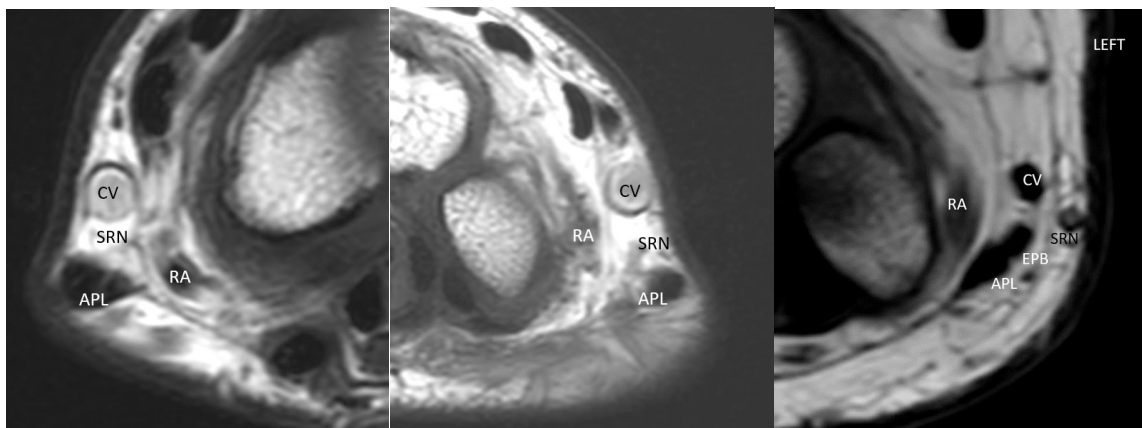
**Table 1:** MRI protocol. TE – Echo time, TR – repetition time, NA – number of averages, AT – acquisition time

	TE [ms]	TR [ms]	NA	Slice thickness [mm]	Interslice gap [mm]	AT [min]
Forearm coronal	15	280	2	3.5	0.4	1:16
Forearm axial	15	609	2	3	0.3	3:50
Wrist coronal	22	500	3	3	0.3	3:21
Wrist axial	24	540	2	3	0.3	3:41

**Figure 1.** Hands of the student on the keyboard. Despite a position close to neutral a substantial abduction of the first metacarpal bones and limited extension of the first metacarpophalangeal joints can be observed.



**Figure 2.** MRI sections at the level of radiocarpal articulation of the right limb (left most picture) and the left limb (middle picture) show bilaterally a sole tendon of abductor pollicis longus in the first extensor compartments. A corresponding section through a normal left wrist of another patient is shown for the reference (right most picture). APL – abductor pollicis longus, EPB – extensor pollicis brevis, CV – cephalic vein, SRN – superficial branch of the radial nerve, RA – radial artery.



**Figure 3.** MRI sections through distal forearms. Absent bellies of EPB muscles. APL – abductor pollicis longus, EPL – extensor pollicis longus.

