

# A cluster of dysmorphologies in a male human body: The value of anatomical variants in health sciences student training

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## SUMMARY

The human body is known to contain many variations in its normal structure which, while of interest to teachers of anatomy, may be vexing to health sciences students when compared to the description of “normal” anatomy in their textbooks. However, these variations, and even dysmorphologies, pose interesting and sometimes challenging learning experiences to students during dissection of the body. Such an instance occurred for undergraduate medical students in the School of Anatomical Sciences, University of the Witwatersrand, South Africa, when three unrelated dysmorphologies were discovered while undertaking a full dissection of a donor’s body. An aberrant right subclavian artery was found in the thoracic cavity and two further dysmorphologies, a supernumerary kidney and accessory indentations on the diaphragmatic surface of the liver presented on dissection of the abdominal cavity. The aberrant right subclavian conformed with previous descriptions of the anomaly. However, the supernumerary kidney lacked a ureter, was lobulated and contained large blood-filled spaces, with histological evidence of urinary tubules in the intervening connective tissue. The accesso-

ry hepatic indentations varied in depth, with the deeper one forming a fissure and the less deep indentation, a sulcus. While the described dysmorphologies vary in their incidence, the occurrence of a cluster of three within one body provided a significant opportunity for the students to review the normal anatomy, and especially the complex development of the structures, as well as the clinical significance of each.

**Key words:** Accessory hepatic indentations – Dysmorphologies – Morphological anatomy – Right aberrant subclavian artery – Supernumerary kidney

## INTRODUCTION

The morphology of the human body is complex and may be further complicated by variations in the arrangement and/or position of organs and other structures. While variations are not considered to be abnormal (Willan and Humpherson, 1999), they present a distinctive learning perspective for the health sciences student from that

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depicted in anatomical textbooks and demonstrate the uniqueness of each individual. Variations of structures may include dysmorphologies, which are expressions of human teratology or alterations of normal morphology (Merriam-Webster; Richtsmeier, 2017) of congenital origin. When these dysmorphologies occur, they provide the student with a unique atlas within each body and an irreplaceable learning experience. Variations and dysmorphologies discovered during the dissection of the human body furnish students with insight into what they may stumble upon in clinical practice. While variations and dysmorphologies may generally not affect the function of the relevant organ (Kachlík et al., 2020), their identification may play a significant role in diagnosis as well as minimising misdiagnosis in clinical practice.

A unique learning experience arose for a group of medical students in their second year of study while undertaking full body dissection of a donor body as part of their anatomy course in the School of Anatomical Sciences, Faculty of Health Sciences, University of the Witwatersrand, South Africa. Three major dysmorphologies were noted during the dissection. One of these abnormalities occurred in the thoracic cavity, while the remaining two were located in the abdominal cavity. In the thoracic cavity the absence of the brachiocephalic trunk as a major branch of the aortic arch was accompanied by an aberrant right subclavian artery as the distal-most branch of the aortic arch. In the abdomen, a third (supernumerary) kidney without a ureter was present on the left side. Finally, two accessory hepatic indentations were noted on the diaphragmatic surface of the liver.

This study thus describes three unusual dysmorphologies occurring together in one individual and reviews their development and clinical relevance as pertinent to variations which may occur in the human body.

## MATERIALS AND METHODS

Permission to undertake the study was provided by the Collections Committee of the School of Anatomical Sciences, University of the Witwatersrand, and by an ethics waiver from the Human

Research Ethics Committee (Medical), University of the Witwatersrand (W-CBP-220504-01).

A South African white male presented for dissection. The individual – aged 74 years – had donated his body for teaching and learning to the School of Anatomical Sciences, Faculty of Health Sciences, University of the Witwatersrand. The cause of death of the individual as noted on the death certificate listed “cutaneous squamous cell carcinoma with metastases in the brain, liver, lung”. A secondary diagnosis was also provided which stipulated: “brain (occipital and parietal area), numerous pulmonary nodules, liver”.

The dissection of the body was carried out in full as per the instructions detailed in the practical manual “Practical Anatomy” (Kieser and Allan, 2020). The length, breadth, and depth of the kidneys as well as the length and depth of the liver indentations were measured using a manual sliding vernier scale calliper with an accuracy of 0.05mm. Photos of the morphological structures of interest were captured using a Nikon DSLR D90 camera.

Segments of the supernumerary kidney were removed for histological investigation and fixed in 10% neutral buffered formalin for 48 hours. Following fixation, the tissue was routinely dehydrated through a graded series of alcohols, cleared in xylene, and embedded in wax. Sections of the wax-embedded tissue were cut on a microtome at 5µm and placed on silane-coated glass slides. The sections were then rehydrated through a graded series of alcohols and stained with haematoxylin and eosin, and following cover-slip-ping with Entellan®, were examined with a Zeiss (Primo Star) compound microscope to allow for a general appraisal of the tissue. Following analysis by a histologist, the sections were independently appraised by a pathologist, and the diagnosis was confirmed.

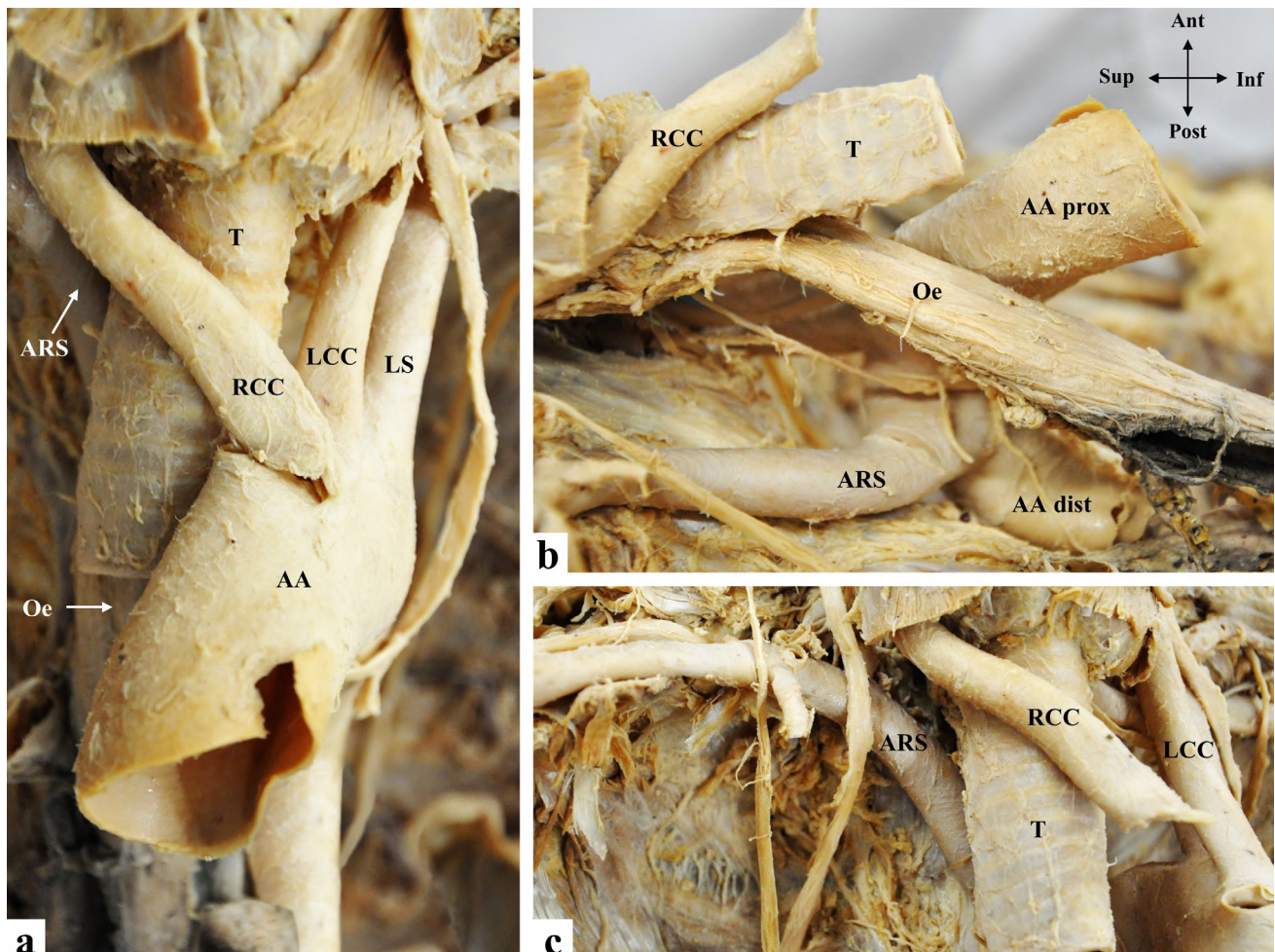
## RESULTS

The three major abnormalities found during the dissection of the body included an aberrant right subclavian artery, a supernumerary kidney, and accessory hepatic indentations. Each structure presented with distinct and clearly divergent morphology from the expected anatomy.

### Aberrant Right Subclavian Artery

Generally, three major arteries arise from the aortic arch: the brachiocephalic trunk (which branches into the right subclavian artery and right common carotid artery), the left common carotid artery, and finally, the left subclavian artery. In the male body dissected in this study, four major branches arising from the aortic arch were observed. The brachiocephalic trunk was absent and instead, four large arteries arose directly from the aortic arch. These were (from proximal to distal) the right common carotid artery, the left common carotid artery, the left subclavian artery, and most distally, the right aberrant subclavian artery (Figs. 1a, b, c).

The right common carotid artery, which originated most proximally on the superior aspect of the arch, continued in a right superolateral direction, passing anterior to the trachea at the level of T3. The artery then passed along the right inferior margin of the thyroid gland and continued superiorly, parallel to the larynx. The next branch arising from the aortic arch was the left common carotid artery, which originated from the superior aspect of the aortic arch and continued superiorly along the left margin of the thyroid gland and travelled parallel to the larynx. Next, the left subclavian artery originated from the left superolateral aspect of the aortic arch and passed superolaterally, posterior to the left anterior scalene muscle. There-



**Fig. 1.-** Photographic representation of right aberrant subclavian artery. **1a:** Anterior view of the aortic arch (AA) showing its four major branches: Right common carotid artery (RCC), left common carotid artery (LCC), left subclavian artery (LS), and the aberrant right subclavian artery (ARS) running posterior to the trachea (T) and oesophagus (Oe). **1b:** Right lateral view showing the origin of the aberrant right subclavian artery (ARS) from the aortic arch (AA). Proximal (AA prox) and distal (AA dist) parts of the arch are indicated. The right common carotid artery has been separated from the aorta. Note the position and course of the aberrant right subclavian posterior to the trachea (T) and oesophagus (Oe). **1c:** Anterior view of the superior thoracic region displaying the course of the aberrant right subclavian artery (ARS) posterior to the trachea (T) (oesophagus cannot be seen in this view). The aortic arch has been reflected out of view.



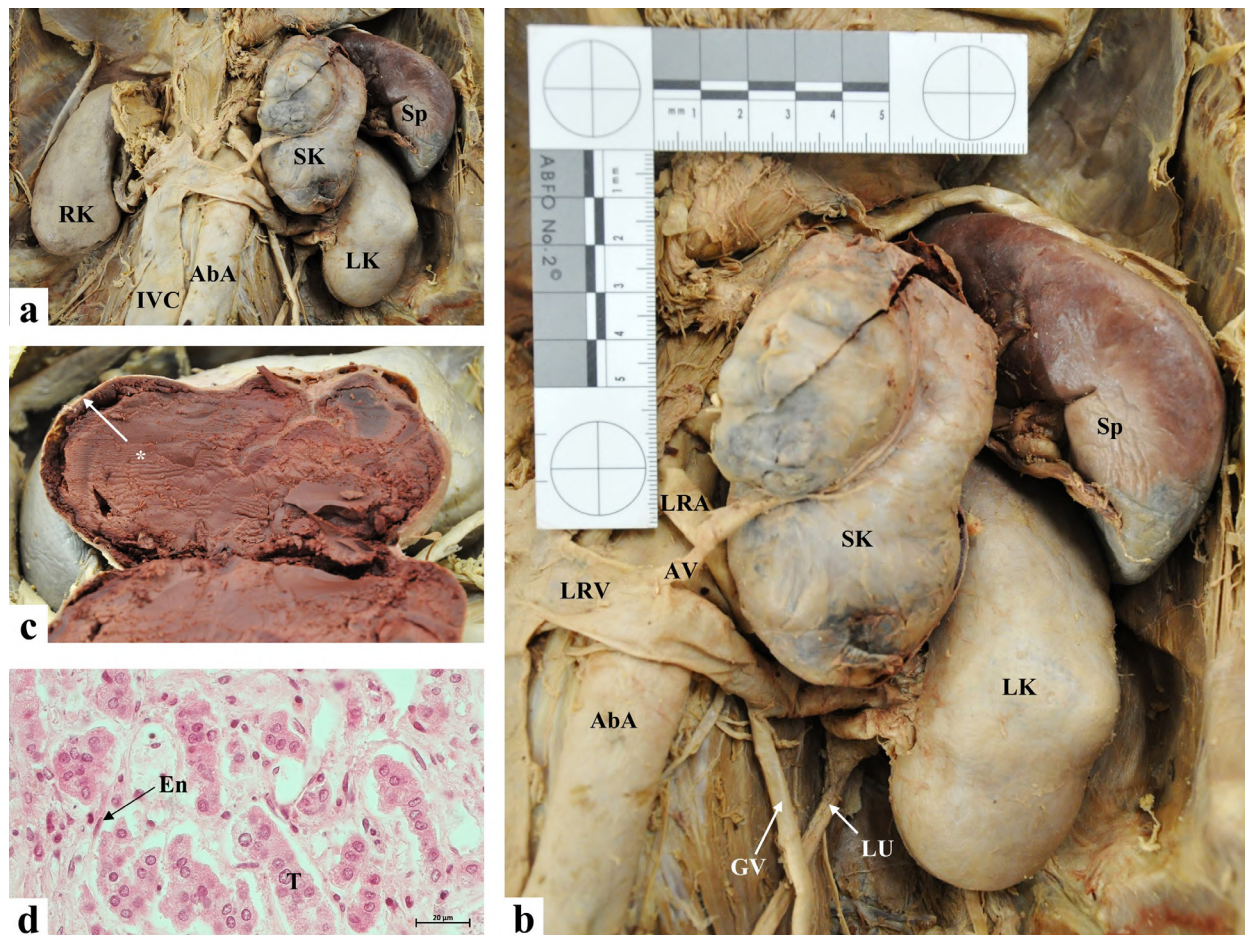
after, it continued laterally to the external border of the first rib, where it became the left axillary artery as expected.

The aberrant right subclavian artery arose from the aortic arch as the distal-most branch, where it originated from the posteromedial aspect (Fig. 1b). It continued in a right superomedial direction, passing posterior to the oesophagus at the level of T3, then superolaterally to the external border of the first rib and continued into the right upper limb (Fig. 1c).

### Supernumerary Kidney

Two normal kidneys plus a third ectopically-placed kidney were found in the body upon

dissection. The left and right normal kidneys occurred retroperitoneally against the posterior abdominal wall in the left and right lumbar regions, respectively. The normal kidneys occupied an oblique position, had a smooth surface and were ovoid in shape, lying on either side of the vertebral column between the levels of T12 and L3. As expected, the right kidney occurred more inferiorly than the left within these vertebral levels due to the presence of the liver in the upper right quadrant of the abdomen. A left and a right ureter, respectively, were found originating from each kidney. These too were retroperitoneal and ran inferiorly after exiting each renal hilum, then passed over the bifurcation of the common iliac arteries and entered the bladder along the later-



**Fig. 2.-** Photographic representation of the supernumerary kidney. **2a:** Anterior view of the abdominal retroperitoneal organs in situ including the supernumerary kidney to show the anatomical arrangement of the two normal kidneys and the supernumerary kidney (SK). The liver, stomach, and intestines have been removed. The diaphragm has been reflected. IVC – Inferior vena cava; RK – Right kidney; LK – left kidney; AbA – abdominal aorta. **2b:** Anterior view of the left retroperitoneal region of the abdomen, including the supernumerary kidney (SK) and its accessory vein (AV) – a tributary of the left renal vein (LRV). Note the lobulated appearance of the supernumerary kidney compared to the normal left kidney (LK). Sp – Spleen; LU – Left ureter of normal kidney; GV – Gonadal (testicular) vein; AbA – Abdominal aorta; LRA – Left renal artery. Note the absence of a ureter and/or renal pelvis in relation to the supernumerary kidney. **2c:** A sagittal section of the supernumerary kidney showing the renal capsule (\*) and blood-filled cavities. **2d:** Histological section of the supernumerary kidney showing tubules reminiscent of the nephric loops. T – Tubules; En – Endothelium of a peritubular blood vessel. Light microscopy (x40), scale bar = 20 µm.



al wall of the pelvis. The left and right suprarenal glands were topologically normal and their anatomical relations with each respective kidney were typical.

A third, supernumerary kidney (Figs. 2a, b) was found anteromedial to the left kidney and was contained in its own capsule (Fig. 2c) with its longitudinal axis in the same plane as the longitudinal axis of the left kidney. It occupied a region posterior to the stomach, pancreas, and jejunum. The inferior margin of the supernumerary kidney was positioned more superiorly than the inferior margin of the left kidney. While the structure was retroperitoneal, a large protuberance which occurred on the anterior aspect of the supernumerary kidney, bulged the peritoneal lining.

The supernumerary kidney resembled the basic ovoid shape of a normal kidney, but its surface was lobulated, reminiscent of its embryonic state. It was 103 mm in length (greatest length mea-

sured along its longitudinal axis; compared with the normal kidneys: R = 113 mm; L = 117 mm), 57 mm wide (R = L = 52 mm), and with an anteroposterior depth of 48 mm (R = 39 mm; L = 45 mm).

While a slight indentation reminiscent of a hilum was noted on the supernumerary kidney, there was no evidence of typical hilar structures, namely a renal pelvis, calyces, or a ureter, nor any evidence of tearing of the latter had it been present. This was further confirmed on sectioning. The supernumerary kidney was drained by a small tributary of the left renal vein. This tributary exited the supernumerary kidney on its anterior surface. The origin of the supernumerary kidney's arterial supply, however, is unclear and is likely to have been inadvertently severed during dissection.

Large blood-filled spaces were evident in the histological sections of the tissue, but no glomeruli nor collecting ducts were found. Connective tissue



**Fig. 3.-** Diaphragmatic surface of the liver showing the deep accessory hepatic fissure (AHF) on the left, and the more superficial accessory hepatic sulcus (AHS) to the right.

strands occurred between the blood-filled spaces and contained blood vessels and tubules reminiscent of the nephric loops (Fig. 2d). The tissue was confirmed by a pathologist to be nephric tissue.

### Accessory Hepatic Fissure and Sulcus

In addition to the major hepatic fissures which were evident on dissection, two accessory indentations were noted on the diaphragmatic surface of the liver of the dissected body. These indentations were respectively termed as a fissure, due to its depth, and the other a sulcus, as it was more superficial/lacked depth. The deeper left accessory indentation (fissure) measured 77 mm in length, 4.5 mm average width, and extended into the parenchyma to a depth of 14 mm. The right depression (sulcus) was shallow and appeared to be slightly longer than the fissure. Both indentations ran in a supero-inferior direction – parallel to each other (Fig. 3). The respective midlines of the accessory hepatic fissure and sulcus were separated by 29 mm. No supernumerary/aberrant ribs were present, and no hypertrophied bands of diaphragmatic muscle were found.

## DISCUSSION

The occurrence of multiple dysmorphologies in a human body present with an opportunity for students and staff in anatomy to discover the wide anatomical variation that can occur. These manifestations also better prepare students for their future role as clinicians, when variations and abnormalities may present unexpectedly in their patients – especially during surgery. Any variations and dysmorphologies found during dissection are therefore an excellent learning experience and should be considered carefully in preparation for making diagnoses and developing treatment options in future patients.

### Aberrant Right Subclavian Artery

The fourth (aberrant) branch of the aortic arch observed in this dissection resembles the anatomy previously described as *arteria lusoria* (Molz and Burri, 1978; Polguy et al., 2014) or *arteria subclavia aberrans retrooesophagus* (Kachlik et al., 2020). This congenital abnormality has a female predominance (Molz and Burri, 1978; Polguy et

al., 2014) and is the most common embryological abnormality of the aortic arch (Darwazah et al., 2015; Polguy et al., 2014), having an incidence ranging between 0.16-4.4% (Freed and Low, 1997; Natsis et al., 2017; Ramaswamy et al., 2008; Rosa et al., 2003). This abnormality is said to have been first described by Hunauld in 1735 (Freed and Low, 1997).

Developmentally, the aortic sac gives rise to the pharyngeal arch arteries (Allan and Kramer, 2009). Six pairs of bilateral arteries leave the aortic sac and pass through each pharyngeal arch before joining the dorsal aorta. However, the number of arch arteries is dynamic, as the pharyngeal arches develop and regress at various stages (Benson et al., 1992), with the fifth arch being rudimentary.

The right and left dorsal aortae receive the pharyngeal arch arteries bilaterally. The dorsal aortae remain paired at the level of the pharyngeal arches, but merge caudally to create the descending aorta (Rosen and Bordoni, 2022). Seven bilateral cervical intersegmental arteries originate from the dorsal aortae. The left subclavian artery arises entirely from the left seventh intersegmental artery, while the right subclavian artery normally arises from the right fourth pharyngeal arch artery proximally, and the right seventh intersegmental artery distally (Rosen and Bordoni, 2022).

An aberrant right subclavian artery results when the right fourth pharyngeal arch artery regresses, leading to the loss of the right subclavian's typical proximal section (Polguy et al., 2014). Consequentially, the seventh right segmental artery serves as the sole source of the artery resulting in an independent branch originating from the aorta and an independent origin for the right common carotid artery. In 60% of cases of aberrant right subclavian artery, an aortic diverticulum known as Kommerell's diverticulum is present (Domínguez-Massa et al., 2019), which is a fragment of the right aortic arch present at the origin of the aberrant right subclavian artery (Domínguez-Massa et al., 2019; Freed and Low, 1997).

Individuals with an aberrant right subclavian artery may be asymptomatic, and are often dis-

covered accidentally (Darwazah et al., 2015). In cases where the aberrant artery compresses the oesophagus, thereby creating a physiological constriction, dysphagia lusoria may occur (Polgaj et al., 2014), which is of clinical significance. However, the aberrant right subclavian artery presents with symptoms in only 7-10% of adult patients (Delap et al., 2000) and thus must be considered during surgical procedures involving the oesophagus (Mahmodlou et al., 2014). In patients in which progressive dysphagia presents, it is imperative to be aware of the possible courses of the aberrant artery such as the usual retro-oesophageal course or the uncommon route anterior to the trachea or oesophagus, ensuring an effective and encompassing surgical approach. Furthermore, anomalies such as non-recurrent right inferior laryngeal nerve, right sided aortic arch and a common origin of the common carotid arteries have been reported in association with an aberrant right subclavian artery (Epstein and DeBord, 2002).

### **Supernumerary Kidney**

While renal abnormalities are not uncommon, a supernumerary kidney is rare (Kumar et al., 2019; Mejia et al., 2018; Sureka et al., 2013) with less than 100 cases said to have been reported in the literature (Ardalan, 2016; Sureka et al., 2013). The first case was described by Martius in 1656 (Krakhotkin et al., 2021). The incidence of supernumerary kidney has not been calculated due to its infrequent appearance (Tada et al., 1981), although the introduction of CT scans and MRI are now likely to document this variant more frequently. Most supernumerary kidneys are said to be located on the left-hand side (Krakhotkin et al., 2021; Tada et al., 1981) and are equally distributed in males and females (Ardalan, 2016; Tada et al., 1981). However, few cases of a supernumerary kidney without a ureter have been reported in the literature (Ardalan, 2016; Gray and Skandalakis, 1972).

During human development, a mesonephros develops prior to the formation of the normal adult kidney (metanephros) and undergoes an almost identical development to that of the metanephros (Allan and Kramer, 2009). Two important constit-

uents are present during the normal development of both the mesonephric and the metanephric kidney, the ureteric bud (from the mesonephric duct) and the nephrogenic cord (Allan and Kramer, 2009). The metanephrogenic blastema, which arises from a condensation of nephrogenic cord mesoderm, induces the cells of the ureteric bud to differentiate and develop the renal collecting system. Simultaneously, the ureteric bud induces the metanephrogenic blastema to differentiate into the renal excretory system, consisting of the renal corpuscle, the nephric loops, and the proximal and distal convoluted tubules. The collecting duct system, derived from the ureteric bud, develops into the collecting ducts, major and minor calyces, renal pelvis, and the ureter. While one of the earliest studies of kidney development by Felix (1911) provided information on the steps of the development of the nephron in the mesonephros, the information was based mainly on comparative embryology and described mesoureterers originating from the mesonephric (Wolffian) ducts. Recently, Landsman and Ludwig (2005) through their study of serial sections of human embryos have definitively shown that the cone of the mesonephric duct becomes the mesoureter (and subsequently the ureter of the adult kidney).

Initially, the metanephric (adult) kidney develops in the pelvic region, but progressively ascends until each kidney encounters the suprarenal gland. During the ascent of the kidneys, the hilum of each kidney rotates from initially facing anteriorly to finally facing medially in the adult. Thus, all nephric hilar structures normally exit/enter the kidney on its medial surface. In the case of the supernumerary kidney found in this study, an accessory renal vein exited the supernumerary kidney anteriorly, perhaps indicating that the normal rotation did not occur.

During the developmental ascent of the kidneys, their blood supply progressively changes. Initially the artery to each respective kidney originates from the corresponding common iliac artery (Allan and Kramer, 2009). With cranial migration of each kidney, the early blood supply degenerates and is replaced by intermediate renal arteries that originate from the distal part of the aorta. When the kidneys finally encounter the supraren-

nal glands, cranial migration of the kidney ceases, the intermediate blood supply degenerates, and the final arterial supply (adult renal artery) originates from the abdominal aorta. In some cases, these intermediate vessels fail to degenerate and present as accessory renal arteries or veins (Gupta et al., 2012).

The lobulated appearance of the dissected kidney, while reminiscent of an embryonic kidney that naturally smooths out due to growth of the nephrons over time (Shanthi D'Sa et al., 2022), is believed to be due to the masses of blood-filled spaces. Tubules within the connective tissue strands of the supernumerary kidney confirmed the diagnosis of a kidney by a histopathologist.

The supernumerary kidney found upon dissection in this study may be the result of the abnormal division of the nephrogenic cord into two metanephrogenic blastemata (Kumar et al., 2019; N'Guessan and Stephens, 1983) with or without division of the ureteric bud or a splitting of the ureteric bud (Ardalan, 2016). Generally, the supernumerary kidney may have a partially or completely duplicated ureter (N'Guessan and Stephens, 1983). However, it cannot be determined whether the supernumerary kidney found in this study developed *ab initio* or resulted from the splitting of the nephrogenic cord into two metanephrogenic blastemata (Kumar et al., 2019; N'Guessan and Stephens, 1983).

A supernumerary kidney on the left side was reported in a 45-year-old male in 1911 (Dixon, 1911). In this case, however, the supernumerary kidney presented with a distinct ureter. Few cases of supernumerary kidneys without ureters have been described in the literature although reference to detached masses of metanephrogenic tissue without ureters have been reported and are said to undergo differentiation. These masses were designated as "beinieren" by Neckar-Sulmer (1914) (see Gray and Skandalakis, 1972). It is highly probable that the supernumerary kidney found in the dissected body was not functional during the individual's lifetime and is likely to have been asymptomatic (Ardalan, 2016).

Clinically, upon analysis and interpretation of various non-invasive imaging techniques, super-

numerary kidneys may be misdiagnosed. Rehder et al. (2019) suggest that the primary reason for an initial incorrect diagnosis of supernumerary kidney in 78% of the cases studied may be attributed to the relatively unfamiliar nature of a supernumerary kidney. Furthermore, supernumerary kidney is not regarded as a differential diagnosis. Rehder et al. (2019) further argue that there is currently no universally accepted approach to diagnose supernumerary kidneys consistently and accurately. Two major reasons for the importance of detecting and correctly interpreting cases involving a supernumerary kidney are provided by Rehder et al. (2019), including the importance of preventing the performance of unnecessary procedures, and more importantly, avoiding complications of superfluous surgical procedures as a result of incorrect diagnoses.

#### **Accessory Hepatic Fissure and Sulcus**

The topography of the accessory hepatic fissure and sulcus found in the dissected body is consistent with those described by Macchi et al. (2003) and are sometimes termed "cough furrows" (Nayak et al., 2017). Zahn (1882) as cited by Macchi et al. (2003) most likely provided the earliest description of accessory fissures of the liver. These indentations are said to occur frequently (Auh et al., 1984; Ono et al., 2000).

"Weak zones" that correspond to the margins between terminal branches of adjacent segmental portal veins may be the origin of hepatic indentations (Macchi et al., 2003). These weak zones in the superficial hepatic parenchyma are said to be particularly impressionable, and thus, as a result of pressure from the diaphragm on the liver, cause the formation of hepatic sulci or fissures (Macchi et al., 2003). In certain pathologies (such as the metastases to the lung found in the dissected body) that prompt a chronic increase in diaphragmatic activity, the increased pressure acts mainly at the preformed weak zones, which results in the portal fissures or "cough furrows" (Macchi et al., 2003).

Large variations in the incidence of accessory fissures on the diaphragmatic surface of the liver have been reported, ranging from 2% (Saritha et al., 2015) to 63% (Auh et al., 1984). The notice-



able discrepancy in incidence could be attributed to factors that may lead to the formation of these accessory hepatic indentations on the weaker regions of the liver, for example pressure from the ribs or the diaphragmatic muscle (Reddy et al., 2016). An understanding of these indentations and their causes may be of significance in the precise diagnosis and interpretation of sonography and/or CT imaging (Auh et al., 1984), as these indentations may be confused with hepatic cysts or haematomae. Knowledge of the presence of these indentations is of importance in preparation for hepatic segmental resections and transplantation procedures (Reddy et al., 2016).

### Limitations of the study

Medical records of the donor body are not available to the authors. The study was limited by the unavailability of photographic records of rare supernumerary kidneys without a ureter, with which the present case could be compared.

### CONCLUSION

Dysmorphologies found in the human body during dissection such as those described in this article are of vast importance in that they add to knowledge on human variability, allow for in-depth discussion of their embryological development and provide a clinical focus and basis for studying morphological anatomy. In addition, when variants of this nature are discovered by health sciences students themselves during dissection, it provides a vital lesson of the uniqueness of each human body. This experience will remain with the students far into their clinical years and may remind them of the difficulties in, and importance of, diagnosing structures clearly before surgical intervention is contemplated.

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