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CASE REPORT

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Coexistence of Left Internal Carotid Agenesis, Klippel-Feil Syndrome and Postaxial Polydactyly

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Summary

Background: Internal carotid artery agenesis is a rare anomaly that can be clinically asymptomatic. Klippel-Feil syndrome is a skeletal malformation characterized by vertebral fusion. Presence of postaxial polydactyly is suggestive of an underlying syndrome.

Case Report: We report a rare case of a 44-year-old patient with non-specific symptoms and an association between these three rare abnormalities. Vascular anomalies were found using intracranial MR angiography and multi-detector CT angiography of the supraaortic arteries.

Conclusions: Presence of a single aforementioned anomaly requires cautious imaging assessment in order to detect possible associated anomalies and avoid diagnostic pitfalls. A possible common genetic background could explain the coexistence of these three anomalies.

MeSH Keywords: Angiography • Carotid Artery, Internal • Klippel-Feil Syndrome • Polydactyly

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Background

Isolated agenesis of the internal carotid artery (ICA) is an uncommon anomaly with low incidence, occurring in less than 0.01% of the population [1] and mostly not accompanied by other abnormalities. Klippel-Feil syndrome (KFS) is a congenital skeletal anomaly with incidence of up to 0.5% [2] that can be combined with additional physical abnormalities. Postaxial polydactyly (PAP) is extremely rare as an isolated anomaly and usually appears as a part of a syndrome. Herein, we present a case of a 44-year-old female patient with non-specific symptoms and with an association between these three rare abnormalities. Concomitance of these three anomalies has not been reported in the English-language literature to date. The study was approved by the Institutional Review Board and patient informed consent was obtained.

Case Report

A 44-year-old woman presented with a few-year history of neck and lower back pain, as well as a 7-year history of right-sided headaches accompanied by photophobia, phonophobia, nausea and occasional vomiting. She underwent surgical treatment for PAP at the age of 2.

Her neurological status and laboratory tests were within normal range. Brain MR did not detect any parenchymal lesions. However, normal signal void of the left ICA was not detectable (Figure 1). Intracranial MR angiography revealed absence of the left ICA (Figure 2). The left anterior cerebral artery (ACA) and a dilated posterior communicating artery (PCoA) supplied the left brain hemisphere. The left ophthalmic artery originated from the PCoA, while the A2 segment of the left ACA was supplied with blood from the right ACA. CT scan of the temporal bone confirmed absence of the left carotid canal, therefore supporting the finding of left ICA agenesis. Multi-detector CT angiography of the supraaortic arteries revealed hypoplastic left common carotid artery and left vertebral artery (Figure 3). On the same examination we detected fusion of C3-C4 and C7-Th1 vertebral bodies (Figure 4), which was highly suggestive of type II KFS. Abdominal ultrasound was unremarkable.

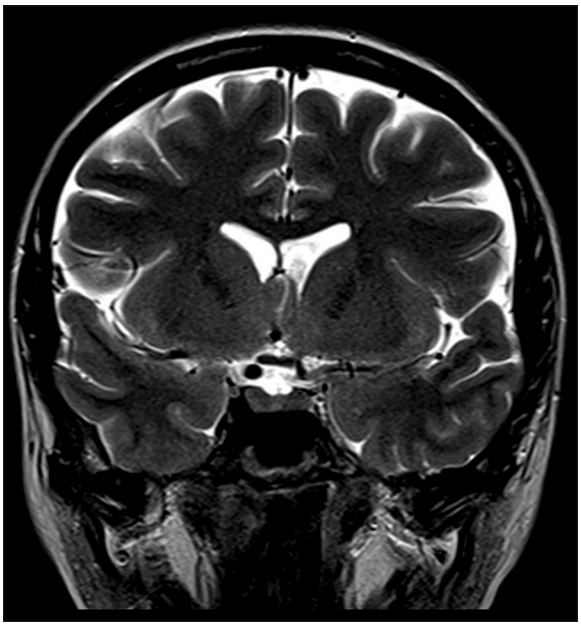


Figure 1. Coronal T2-weighted MRI shows a lack of normal signal void of the left internal carotid artery.

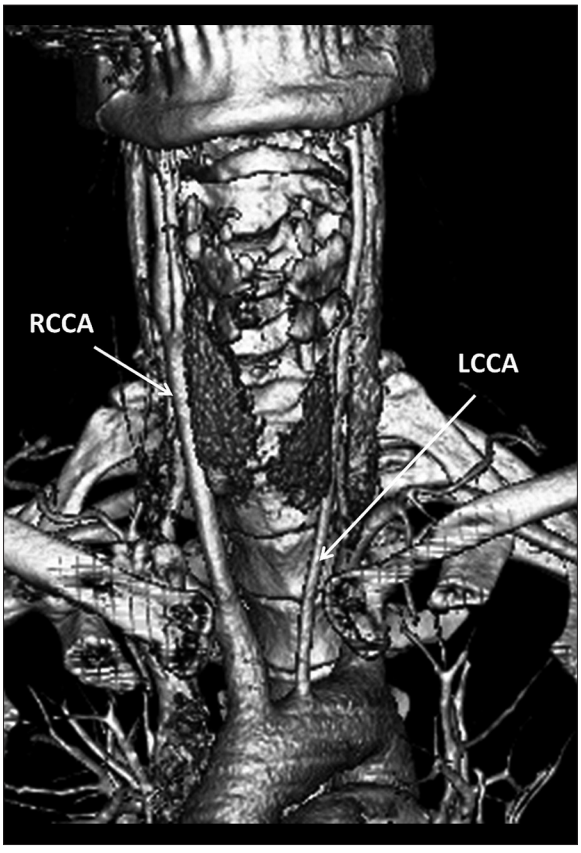


Figure 3. Multi-slice CT angiography of the supraaortic arteries displays a hypoplastic left common carotid artery and agenesis of the left internal carotid artery. RCCA = right common carotid artery, LCCA = left common carotid artery.

Figure 4. Sagittal reconstruction of the cervical spine exhibits fusion of C3-C4 and C7-Th1 vertebral bodies.

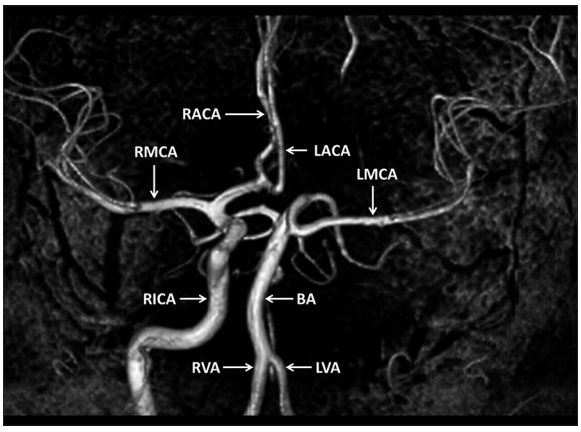
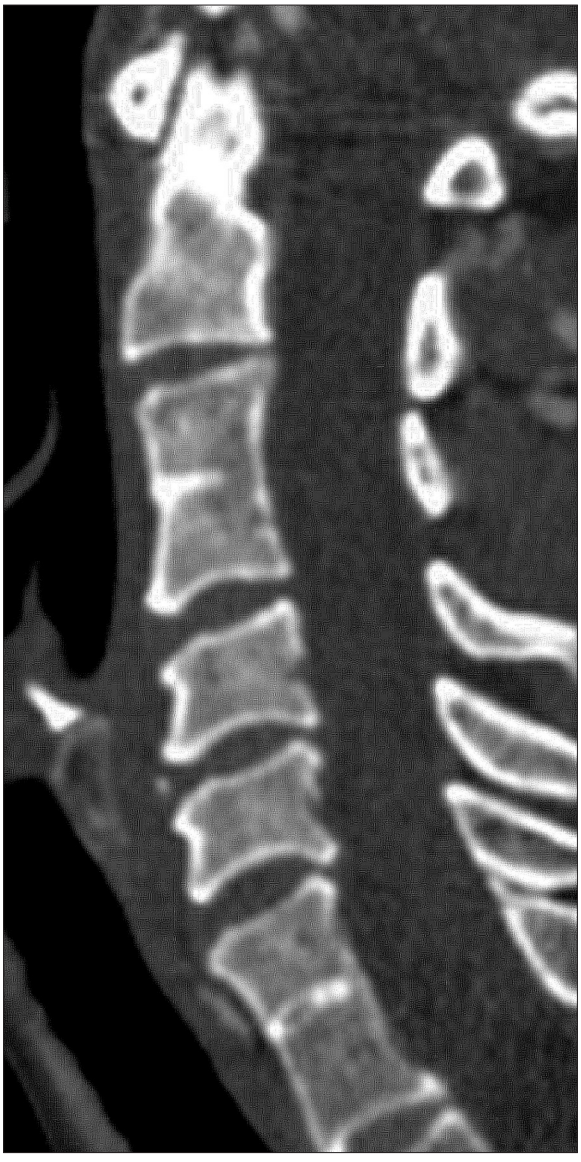


Figure 2. Intracranial MR angiography reveals absence of the left internal carotid artery. RVA = right vertebral artery, LVA = left vertebral artery, BA = basilar artery, RICA = right internal carotid artery, RMCA = right middle cerebral artery, LMCA = left middle cerebral artery, RACA = right anterior cerebral artery, LACA = left anterior cerebral artery.



Discussion

Internal carotid vessels develop early in embryogenesis originating from the third aortic arch and the dorsal aorta, probably as a separate process from embryogenesis of the common carotid and external carotid artery [3]. This patient showed a type A unilateral absence of the ICA [4]. If the collateral circulation provides sufficient blood supply, unilateral ICA agenesis may be clinically asymptomatic. Nonetheless, it can manifest with severe conditions as transient ischemic attacks, cerebral ischemia, subarachnoidal hemorrhage or aneurysms of other intracranial arteries [5]. ICA agenesis is usually an isolated anomaly or associated with aneurysms of the circle of Willis, abnormal arterial collateral channels and dilated vascular channels, cerebral hemiatrophy, Klippel Trenuanay syndrome, arachnoid cyst, as well as with non-cerebral anomalies including cardiac septal defects, tongue hemangioma, angiofibroma with atresia of the upper basilar artery, and neurofibromatosis [5].

KFS is characterized by vertebral fusion, usually at the cervical spine level, as a result of impaired vertebral segmentation. Interruption of early embryonic blood supply in the subclavian arteries, vertebral arteries and/or their branches has been proposed as a possible etiology of KFS [6]. The clinical triad of KFS includes short neck, low neckline and restriction of head and neck movements, but if less than three vertebrae are fused, especially lower cervical vertebrae, restriction of movements is rare. KFS can also be asymptomatic for a long time. More often, KFS is associated with other abnormalities which include musculoskeletal, cardiovascular, urogenital, neurologic, hindbrain, auditory, ocular, craniofacial, otolaryngological, limb and digital anomalies [7]. KFS and ACI agenesis have in common an underlying embryological impairment in vascularization.

PAP as an isolated anomaly in the white population is extremely rare and is indicative of an underlying syndrome. Meckel-Gruber syndrome, Ellis-van Creveld syndrome, chondroectodermal dysplasia, short rib-polydactyly syndrome, Smith-Lemli-Opitz syndrome, trisomy 13, Bardet-Biedl syndrome, McKusick-Kaufman syndrome, orofacial-digital syndromes can include polydactyly [8].

So far, there has been only one report of internal carotid agenesis accompanied by KFS [9]. Isolated KFS and ACI agenesis can exhibit different and possibly severe symptomatology, but can also be asymptomatic. Our patient had a combination of these two anomalies and remained asymptomatic for a long time, presenting later with nonspecific symptoms which can be the manifestation of various illnesses or even of a degenerative spinal disorder. An association between KFS and PAP is extremely rare and has been described in a few cases [10]. PAP has never been reported along with ICA agenesis.

Conclusions

We could hypothesize that those three anomalies may have a common genetic background. Presence of a single aforementioned anomaly requires careful imaging assessment in order to detect possible associated anomalies and avoid diagnostic pitfalls.

Conflict of interest

The authors declare that they have no conflict of interest.

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