A case of Klippel-Feil syndrome with congenital cardiovascular anomalies

Doğumsal kardiyovasküler anomalilerin eşlik ettiği Klippel-Feil sendromu

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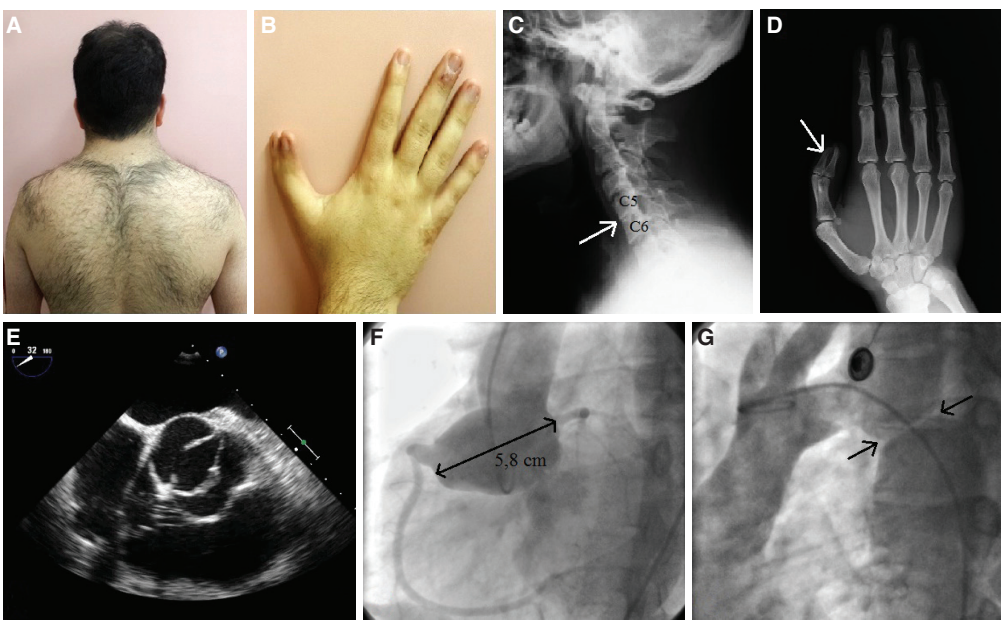
A 21-year-old male patient with no known cardiovascular disease was admitted with hypertension. There was a 50 mmHg blood pressure difference between the two arms. On physical examination; he had short neck stature (Figure A), and cervical limitation of motion and bifid distal phalanx of the right thumb (Figure B). X-ray studies showed fusion of the fifth and sixth cervical vertebrae (Figure C) and bifid distal phalanx of the right thumb (Figure D). Transthoracic and transesophageal echocardiography showed a bicuspid aortic valve (Figure E), an aortic root aneurysm (of sinus valsalva diameter 5.8 cm) and coarctation of the arcus aorta. Subsequent aortography showed an aortic aneurism at the level of sinus valsalva (Figure F), coarctation at the arcus aorta after the left subclavian artery, and post-dilatation of the descendant aorta (Figure G). A 55 mmHg pressure gradient was measured by catheterization between proximal and distal parts of the aortic coartation. Contrast-enhanced thoracoabdominal computed tomography did not reveal any splanchnic organ anomalies. Genetic tests detected the GDF6 genetic mutation, associated with Klippel-Feil Syndrome. Aortic valve replacement with aortic graft implantation were performed, and the patient discharged from hospital on the postoperative 10th day. Klippel-Feil Syndrome was first defined by Maurice Klippel and Andre Feil in 1912. The main characteristics of Klippel-Feil Syndrome are short neck stature, low hairline, and cervical limitation of motion due to fusion of cervical vertebrae. Scoliosis, spina bifida, wing scapula, extremity anomalies, cleft palate, and renal and cardiovascular anomalies may accompany these. Precise diagnosis is based on genetic tests. Potential cardiovascular anomalies should be kept in mind when dealing with skeleton anomalies in patients with Klippel-Feil Syndrome.



Figures– (A) Short neck stature **(A)** and bifid distal phalanx of the right thumb **(B)** in a patient with Klippel-Feil Syndrome. X-ray showing fusion of fifth and sixth cervical vertebrae **(C)** and bifid distal phalanx of the right thumb **(D)**. Transesophageal echocardiography showing the bicuspid aortic valve **(E)** with aortic root dilatation. Aortography showing the aortic aneurysm at the level of the sinus valsalva **(F)**, coarctation of the arcus aorta after the left subclavian artery and **(G)** post-dilatation of the descending aorta.