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Case Report

Navigating challenges: Cleidocranial dysplasia and complexities in transvenous pacemaker implantation ☆

Hanaa El Ghiati, MD, Abdelilah Benelmakki, MD, Jihane Fagouri, MD, Jaouad Nguadi, MD, Meryem Bennani, MD, Jamal Kheyi, MD, Hicham Bouzelmat, MD, Aatif Benyass, MD, Ali Chaib, MD*

Military Training Hospital of Rabat, Morocco

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ABSTRACT

Cleidocranial dysplasia (CCD) is a rare genetic disorder characterized by skeletal abnormalities, including hypoplastic or absent clavicles, delayed closure of cranial sutures, and dental anomalies.

We present a case of a 72-year-old female with a history of breast cancer treated with mastectomy and radio chemotherapy with the port-a catheter still in place in the left subclavian region. She presented to the emergency room with syncope related to a complete atrioventricular (AV) block. The patient underwent temporary pacing via femoral access while awaiting definitive pacemaker implantation.

The absence of the right clavicle, first observed during prepuncture fluoroscopy and later confirmed on postprocedure imaging, significantly influenced the approach to pacemaker implantation. Venography played a crucial role in visualizing the venous trajectory and guiding the puncture, ensuring successful lead placement.

The procedural challenges encountered due to the patient's skeletal anomalies highlight the importance of individualized approach and careful consideration of anatomical variations in interventional cardiology procedures.

Introduction

Cleidocranial dysplasia (CCD) is a rare genetic disorder characterized by skeletal abnormalities, including hypoplastic or absent clavicles, delayed closure of cranial sutures, and dental anomalies [1]. The condition was first described by Pierre Marie and Paul Sainton in 1898, and it is sometimes referred to as Marie-Sainton syndrome [2].

Permanent pacing is strongly recommended for symptomatic high degree atrioventricular block, and the transvenous pacemaker implantation is considered the conventional

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* Corresponding author.
E-mail address: elghiati.hanaa@gmail.com (H. El Ghiati).

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technique. Accessing the subclavian vein conventionally using fluoroscopy relies on identifying the clavicular bend as a key anatomical landmark for subclavian venous access [3].

Here, we describe a case of a patient who presented with syncope secondary to complete atrioventricular (AV) block necessitating pacemaker implantation, during which we discovered that the patient had cleidocranial dysplasia.

The procedural challenges encountered due to the patient's skeletal anomalies highlight the importance of individualized approach and careful consideration of anatomical variations in interventional cardiology procedures.

Case report

We present a case of a 72-year-old female with a history of breast cancer treated with mastectomy and radio chemotherapy with the port-a catheter still in place in the left subclavian region. She presented to the emergency room with recurrent syncopal episodes. Upon examination, the patient presented an electrocardiogram (EKG) revealed complete atrioventricular (AV) block with low-rate ventricular escape rhythm and high blood pressure. The patient underwent an urgent temporary pacing via femoral access while awaiting definitive pacemaker implantation.

Echocardiography showed a normal left ventricular ejection fraction and no valvular abnormalities.

The patient was then transported to the operating room for permanent transvenous implantation. The absence of the right clavicle, first observed during pre-puncture fluoroscopy and later confirmed on postprocedure imaging, significantly influenced the approach to pacemaker implantation. The right-sided approach for pacemaker insertion was the only option due to the presence of an existing port-a-cath on the left side, precluding a left-sided approach despite the anatomical anomaly of the absent right clavicle.

A blind puncture without clavicle orientation was attempted unsuccessfully. The search for a cephalic vein was also tried but did not succeed. Therefore, a venography was performed by injecting contrast peripherally to visualize the venous trajectory. This technique played a crucial role guiding the puncture, ensuring successful septal ventricular lead placement.

In fact, given the patient's metastatic cancer and low life expectancy, a decision was made to implant a single-chamber pacemaker.

Clinical findings revealed no additional craniofacial features or digit abnormalities, despite the aplastic right clavicle and a sloping right shoulder (Fig. 1). Dentition could not be assessed as all of the patient's teeth had been removed. The patient exhibited short stature but normal intellectual functioning.

A post-procedure chest X-ray confirmed the absence of the right clavicle (Fig. 2), and permitted to rule out a postprocedural pneumothorax. Subsequent CT scan with a 3D bone reconstruction confirmed the diagnosis by showing the absence of the right collar bone; associated with scoliosis (Fig. 3 and 4).

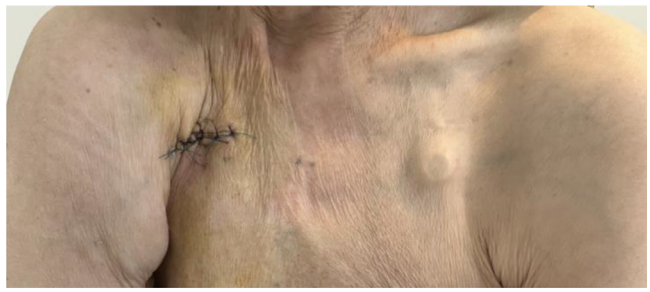

Fig. 1 – Frontal view of the patient showing the absence of the right collarbone comparatively to the left one.

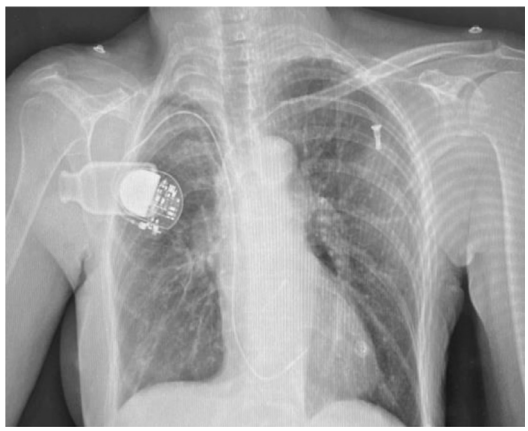

Fig. 2 – Postero-antérior chest X-ray showing the absence of the right clavicle alongside the presence of a port-a-catheter on the left side. It also rules out the presence of pneumothorax after transvenous pacing.

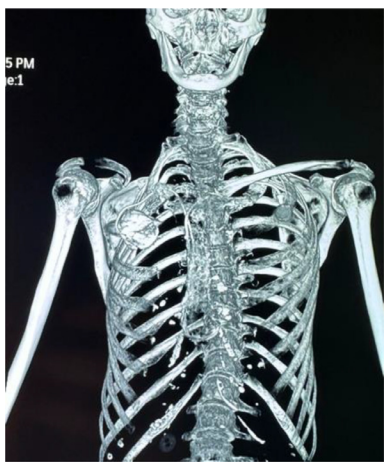

Fig. 3 – Advanced 3D bone reconstruction model showcasing a case of right cleidocranial dysplasia and highlighting the missing collarbone on the right side. It also shows the pacemaker with its ventricular lead on the right, and the port-a-catheter on the left.

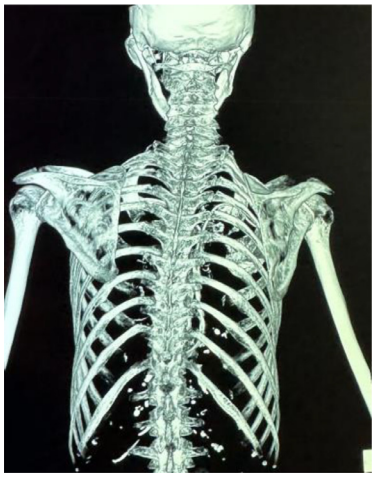

Fig. 4 – Posterior view showing the skull base and spine which appear to be normal.

The patient was discharged 48 hours after implantation, a 6 months follow up showed the patient was in good condition with no complications.

Discussion

The presented case of a 72-year-old female with cleidocranial dysplasia who required transvenous pacemaker implantation due to complete atrioventricular (AV) block poses several intriguing clinical and anatomical considerations.

Cleidocranial dysplasia is a rare congenital skeletal disorder characterized by defective ossification of bones, including the clavicles and cranial sutures, leading to abnormal growth and development of these structures [4]. The skeletal deformity is hypoplasia of clavicle bone, affecting mostly lateral portion [4]. In 10% of cases, clavicle is totally absent. This allows hypermobility of the shoulders resulting in the ability to touch them in front of the chest.

It is an autosomal dominant condition associated with the mutation in the RUNX2 gene which encodes a protein necessary for the correct functioning of osteoblast [5]. This gene is responsible of normal development of human skeleton and guaranties the endochondral ossification. Its mutation is related to the absence of ossification of midline structures and results with various skeletal and dental deformities.

In fact, the primary skeletal deformity observed is hypoplasia of the clavicle, or downright its total absence in 10% of cases. Our patient actually belongs to this 10% as she lacks a right collarbone.

Diagnosis of cleidocranial dysplasia (CCD) is based on clinical and radiographic correlation.

The recent studies are now defining a CCD spectrum disorder that can go from a complete form including cranial dysplasia, hypoplastic or aplastic clavicles and dental abnormalities, to mild or isolated forms. Our patient represented with an isolated case characterized by an aplastic right clavicle.

Placement of a dual-chamber permanent pacemaker is strongly recommended (class I indication) for symptomatic high degree atrioventricular block [3].

The transvenous pacemaker implantation is considered the conventional technique.

Our patient presented with a syncopal third-degree atrioventricular block, but given the patient's metastatic cancer and low life expectancy, a decision was made to implant a single-chamber pacemaker.

Accessing the subclavian vein conventionally using fluoroscopy relies on identifying the clavicular bend as a key anatomical landmark for subclavian venous access. However, in cases where there is clavicular absence or hypoplasia, this landmark becomes unavailable. In such scenarios, alternative radiological and diagnostic techniques become necessary for successful venous access. Our approach involves utilizing intraprocedural venography to achieve successful venous access. This technique has proven to be useful in assisting with the implantation of cardiac electronic devices particularly in cases of venous obstructions [6].

Other strategies for subclavian venous access in patients lacking the clavicular landmark include puncturing the axillary vein using ultrasound guidance [7], performing a cephalic venous cutdown [8], or considering the implantation of a leadless right ventricular pacemaker [9]. In our case, the cephalic vein cutdown was attempted but was unsuccessful.

Clavicular anomalies may also occur following injury to the acromioclavicular joint or from a conservatively managed, displaced clavicular fracture. To our knowledge, there are no other published reports discussing permanent transvenous pacemaker implantation in patients with these specific clavicular disorders. The utilization of intra-procedural venography alongside intraprocedural vascular ultrasound provides a safe and effective approach for accessing the subclavian vein in such cases.

The challenges encountered during this procedure highlight the importance of careful preprocedural assessment and planning in patients with unique anatomical variations such as cleidocranial dysplasia. Factors such as vein anatomy, existing devices or structures, and potential complications associated with altered skeletal morphology must be meticulously considered to ensure procedural success and minimize risks.

Furthermore, the association of cleidocranial dysplasia with cardiac conduction abnormalities, particularly complete AV block in this case is not described. While there is limited literature on the cardiac presentation in patients with this condition, clinicians should be aware of potential fortuitous cardiac associations and tailor management strategies accordingly.

The successful implantation of a single transvenous pacemaker in this patient underscores the adaptability of interventional cardiology techniques in overcoming anatomical challenges. Future considerations for patients with cleidocranial dysplasia requiring cardiac device implantation should include comprehensive pre-procedural imaging, multidisciplinary collaboration involving cardiology and orthopedic specialists, and careful selection of vascular access routes based on individual anatomical variations.

Conclusion

In conclusion, this case highlights the complexities and considerations involved in managing cardiac conduction abnormalities in patients with cleidocranial dysplasia. By integrating advanced imaging modalities and individualized procedural approaches, clinicians can optimize outcomes and ensure safe and effective interventions in this unique patient population.

Patient consent

Consent for publication has been obtained from the patient.

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