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Mitral Regurgitation in Unpublished Mitral Lesions in Cutis Laxa: Case Report and Review of the Literature

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ABSTRACT

Congenital mitral regurgitation (BMI), a rare disease, is defined as any left ventriculo auricular regurgitation due to dysplasia of the congenital mitral valve. It is accompanied by some degree of relative stenosis, and frequently associated with other malformations. Curtis Laxa is a skin disease, either acquired or hereditary, with autosomal and sexual transmission. Redundant skin is often most visible on the neck, hands and groin, but can also be seen on the face, creating an appearance of premature aging. It is associated with severe systemic involvement in some forms. We report a case of severe mitral regurgitation associated with valvular damage in a patient being treated for hereditary Cutis Laxa with other associated systemic damage.

KEYWORDS: Congenital mitral insufficiency (CMI) - Posterior mitral valve hypoplasia (PMVH) - Mitral valve agenesis (MVA) - Cutis Laxa (CL) - CHIARI malformation type 1 (MC1) - Diverticular bladder (DV) - Mitral valve plasty (MVP).

INTRODUCTION

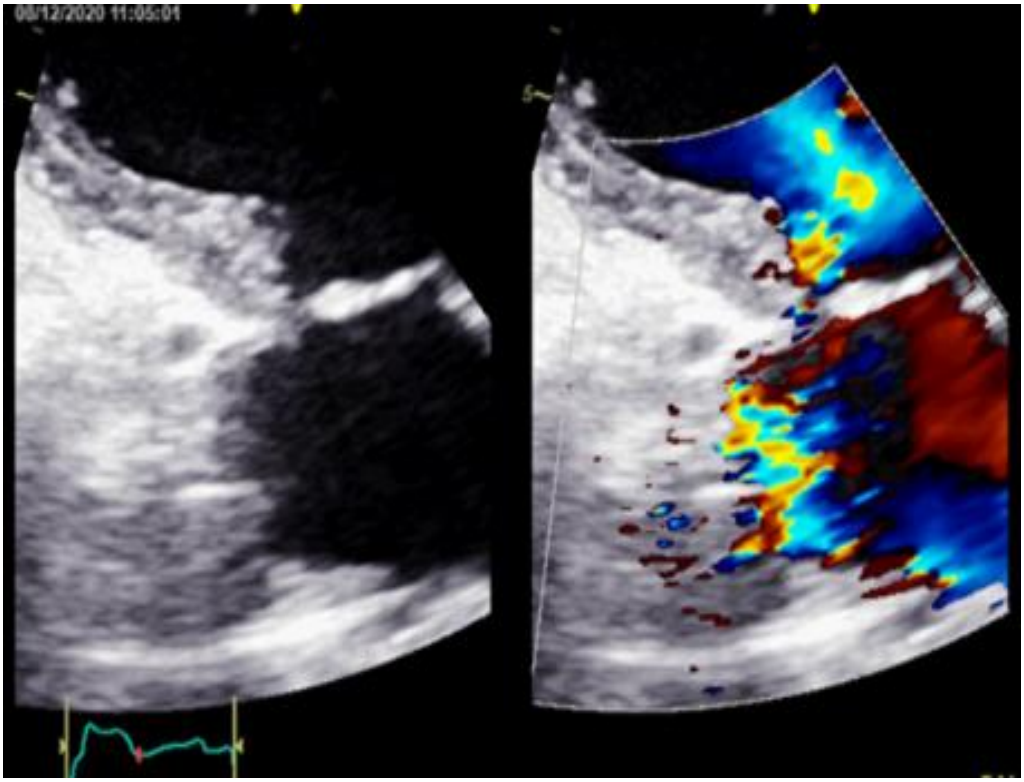
Primary mitral regurgitation results from dysfunction of the mitral valve due to isolated or, more often, joint damage to its component structures by malformation or acquisition. This mitral valve malformation, previously asymptomatic, may become apparent later in adulthood, following its crescendo evolution. It is one of the complications of systemic cardiac damage to the mitral valve of the skin pathology Cutis Laxa. Cutis Laxa is a rare acquired or hereditary disease of elastic tissue, resulting in loose, excessive and hypoelastic skin. Non-dermatological systemic disorders have been reported, including cardiac, neurological, pulmonary, osteoarticular, vascular and urinary tract disorders. In this article, we report the clinical case of a young patient with systemic complications, including severe mitral regurgitation due to valvular damage not described in patients suffering from hereditary Cutis Laxa.

CASE REPORT

A 24-year-old woman, weighing 59 kg and measuring 171 cm, from a 2nd-degree consanguineous parental couple with no family history of malformative disease, from a line of 6 children, 3 of whom were followed for a congenital disease, one of whom, her brother, followed for the same pathology, died in 2016 at the age of 31 year-old . As a child, she was known to have congenital Cutis Laxa (CL) disease, with NYHA class II dyspnoea. At the age of 8, she suffered from rheumatic fever (RF) under injectable extencillin, due to poorly treated recurrent acute tonsillitis (RTA). She has a neurological bladder with self-catheterization on micturition, as well as a documented Chiari malformation type 1 (MC1). Cardiologically, the patient has been followed since 2016 for mitral valve disease with progressive dyspnea. Her infectious history reveals urinary urgency. Her biochemistry was normal and her ECBU was infected on 2 occasions. The electrocardiogram showed regular sinus rhythm, left atrial hypertrophy and complete right bundle-branch block with secondary repolarization disorder . The chest X-ray showed a deformed costal grid with curved ribs, no parenchymal lesions, no signs of pulmonary hypertension, and a double-contour right spine.

Transthoracic echocardiography (TTE) revealed : hypoplasia of the posterior mitral valve responsible for a diastasis leading to significant eccentric mitral regurgitation lining the free wall of the left atrium (LOA) (SOR 0.8 cm2, RV 135 ml). Normal systemic venous return. Generous pulmonary return with dilatation of pulmonary veins. Non-dilated, non-hypertrophied LV, site of relaxation disorders, good global and segmental contractility. LVEF 62%. Ectatic left atrium with SOG :44 cm^2 and SOD :16 cm^2 and undilated LV with good systolic function.

Transesophageal echocardiography (TEE) had revealed : large mitral regurgitation due to hypoplasia of the P2 segment and agenesis of the basal segment of the P3 segment wall with <<compensatory>> hypertrophy of the basal segment of the inferolateral wall. Dystrophic tricuspid valves with ballooning of the posterior valve and moderate tricuspid regurgitation.



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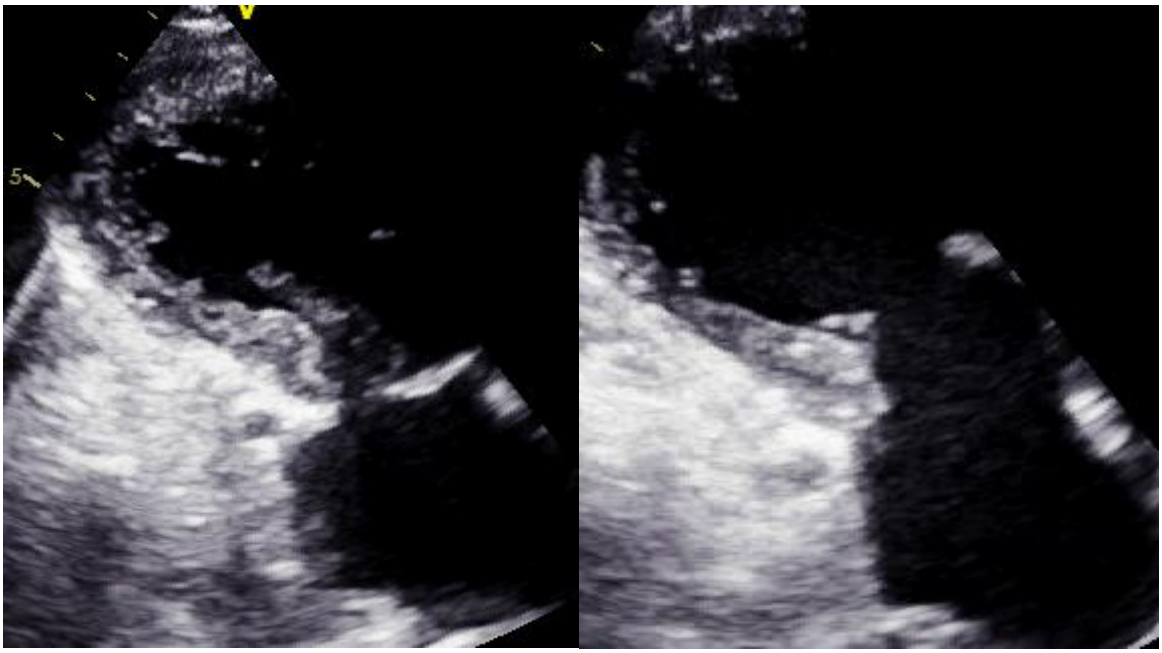

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Figure 1 : Two-dimensional TTE examination : (A) Apical three-chamber view with color doppler : hypoplasia of the posterior mitral valve responsible for a diastasis leading to significant eccentric mitral regurgitation lining the free wall of the left atrium. (B) and (C) Apical three-chamber view : Morphologic examination of the mitral valve : hypoplasia of the P2 segment.

ANATOMICAL OVERVIEW OF THE MITRAL VALVE

The mitral valve apparatus, comprising the mitral annulus, annulus and sub-valvular apparatus (tendon cords and papillary muscles), occupies the left atrioventricular ostium[1].

Mitral veil : this extends around the entire circumference of the mitral orifice. The base of the veil is anchored to a fibromuscular ring. The free edge of the mitral soft palate has several indentations, two of which are constant : the anterolateral (anterior) and posteromedial (posterior) commissures. These

allow division into an anterior valve (large valve) and a posterior valve (small valve) [1]. Each valve has a smooth proximal zone, containing a collagen matrix, and a rough distal zone on which tendinous cords are inserted. Valve coaptation takes place at the level of the roughened zone, the height of this coaptation being around 8 mm on ultrasound. The posterior valve has a greater insertion on the annulus than the anterior valve. The free edge of the posterior valve has two indentations that allow three segments to be identified by notches numbered 1 to 3, from the anterior to the posterior commissure, including a medial segment (P2), two commissural segments, anterior (P1) and posterior (P3). The medial portion is wider than the narrower commissural portions. By analogy, the anterior leaflet is also divided into three segments [1,2].

Mitral annulus : saddle-shaped and flexible. It comprises two major collagenous structures : the right and left fibrous trigones. In front, between the two trigones, the anterior mitral valve is continuous with the aortic valve. The posterior part of the annulus, where the posterior valve is inserted, varies greatly from subject to subject [1]. The shape of the annulus is generally circular in diastole, elliptical in systole, with a reduction in surface area of around 25% for a normal annulus. This systolic contraction plays an active role in valve continence [2].

Subvalvular apparatus (ASV) : this consists of the larger anterolateral and posteromedial papillary muscles, on which the cords are inserted, classified according to their site of insertion into commissural cords, anterior valve cords and posterior valve cords. Valve closure is completed by ventricular pressure exerted on the leaflets, with valve tightness depending on their coaptation surface. The coaptation line lies slightly downstream of the plane of ring [2].

"Primary mitral insufficiency may result from the isolated or, more often, joint dysfunction of the components we have just detailed [2]" .

DISCUSSION AND LITERATURE REVIEW

CL is a disease characterized by wrinkles, redundant, inelastic and sagging skin, caused by defective elastin synthesis or structural abnormalities in the extracellular matrix. The disease can be acquired or hereditary. The hereditary form of cutis laxa (CLH) has an incidence of 1 to 2 cases in 400,000. CLH has 3 innate forms : autosomal dominant, autosomal recessive type I, IIa, IIb, III and X-linked recessive [3,4]. Although all hereditary forms of CL are rare, autosomal recessive Cutis Laxa (ARCL) has been most frequently reported, particularly ARCL-II ; and because of the considerable overlap between these types, precise clinical classification can be difficult [4]. However, based on our patient's family history, her pathology is most likely CLAR.

Systemic involvement has been reported in various forms of CL, predominating in the CLAR form. In a study by D. Bonnet et al [5], cardiac complications were reported in the form of atrio-ventricular valve dysplasia in newborns, including the mitral valve, with no further details on the lesions found. As for L. Baldwin et al, she reports the case of a child with hereditary Cutis Laxa whose color Doppler echocardiography study of the heart showed prolapse of the anterior mitral valve with regurgitation, mild tricuspid regurgitation, and pulmonary hypertension [6].

After research, we realize that no article reports the type of atrio-ventricular mitral cardiac lesions of Cutis Laxa as those found in our patient : hypoplasia of the small mitral valve with agenesis of the basal segment of the P3 segment wall responsible for mitral regurgitation having evolved to the surgical stage. A particular feature of our case is the integrity of his left ventricle, justified by relative stenosis of the mitral valve due to non-dilatation of its annulus, and an ectatic left atrium. There was also associated damage to the tricuspid valve by dystrophy with ballooning, justifying moderate tricuspid insufficiency.

His clinical case is even richer in non-dermatological systemic Cutis Laxa involvement, as it brings together other specific malformations associated with cardiac involvement : Chiari malformation type 1 "which is defined by a herniation of the cerebellar tonsils with walls in the foramen magnum of more than 5mm [7] " until now asymptomatic, a diverticular bladder "protrusions of bladder mucosa through the detrusor muscle fibres. Their walls are devoid of muscular tissue, depriving them of any contractile function, thus inducing urinary stasis [8] ", and a laxity of the right knee.

HPVMP is a rare congenital heart defect, exceptional even outside Cutis Laxa. Characterized by a rudimentary formation of the small valve, it has been described in rare articles, including one by Leili Pourafkari et al in 2018, who described an elongated sail-shaped valve of the anterior mitral leaflet and an immobile hypoplastic small valve formation ; a considerable gap existed between this rudimentary formation and the anterior valve during systole, leading to severe mitral valve regurgitation ; and an associated bicuspid aortic valve anomaly [9]. As for Hakan Ozkan et al, he described the same aspect of the large valve with agenesis of the posterior leaflet [10].. And Claudio G et al showed a cleft in the P2 segment of the small posterior valve, with mitral regurgitation [11].

Furthermore, Bacich Daniela et al revealed an HPVMP, with a primordial chordal apparatus adherent to the endocardium of the posterior wall of the left ventricle (LV), with the anterior leaflet elongated and thickened into meso-tesystolic prolapse of the three scallops and posterior dislocation of the coaptation zone, resulting in an eccentric jet directed posterolateral to the OG. The annulus was slightly enlarged (37 × 38 mm) [12].

All these authors reported an HPVMP or AVM with an anterior mitral leaflet in the shape of an elongated veil, compensating for the lack of valve tightness that led to regurgitation.

The mitral lesions described by these authors are isolated, as in our patient's case, with the exception of the one reported by Leili Pourafkari. However, none of these authors reported a remodeled anterior mitral valve, as their patient had no history of rheumatic fever. And these malformations, although corresponding to those described in our patient, occur without a history of hereditary congenital Cutis Laxa disease, let alone without the context of parental consanguinity, not even

associated with other specific malformations such as MC1 and VD.

MANAGEMENT ISSUES

The patient was presented to a cardiology medical-surgical staff whose indication for mitral plasty was suggested, however, considering :

- The ongoing risk of infective endocarditis of the mitral valve postoperatively via the urinary tract, and taking into account that the patient manually empties her bladder by self-catheterization and has a history of recurrent mictional burning
- A non-dilated LV with preserved systolic function.
- The fairly stationary evolution of her OG dilatation (31 :cm² in 2016 and 42 : cm² in 2020) and her undilated annulus with relative mitral narrowing.

She was rejected for valve surgery, and discharged with a cardiology appointment with ultrasound monitoring every six months.

CONCLUSION

Congenital mitral insufficiency remains a rare malformative disease, regardless of the underlying pathology involved or the context in which it occurs. The occurrence of congenital mitral insufficiency in the systemic attack of a dermatological disease known as Cutis Laxa is rarely reported, if at all. The specific types of mitral valve and neurological lesions described in this article have not yet been described in Cutis Laxa. The prognosis for our patient's longevity is critical in reference to the rare cases reported. And the management of our patient's condition requires not only mitral plasty, but also prevention of the significant ongoing risk of postoperative infective endocarditis. Hence the importance of multidisciplinary management.

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