Transthyretin gene mutations, the most common cause of amyloidosis, lead to neuropathy and often, cardiac disease. Several causal mutations resulting in different phenotypes have been identified.[1]

The mutation found (Val122Ile) produces cardiac amyloidosis in persons older than 60 years with a phenotype similar to that of wild-type transthyretin amyloid, occasionally associated with carpel tunnel syndrome. Between 3% and 4% of black individuals in the United States are heterozygous carriers of this mutation,[2] which is rare in the white population. Although it is considered a rather indolent mutation with late-onset cardiomyopathy, several studies have associated it with greater morbidity and mortality than the wild-type form.[3] When the mutation is homozygous, the risk increases, and heart failure develops earlier and is more severe.[4]

Our patient showed a severe phenotype with rapid progression to heart failure, New York Heart Association functional class II/IV, left ventricular dysfunction, and considerable elevation of N-terminal pro-brain natriuretic peptide.

Genetic study was not only useful for the diagnosis of hereditary amyloidosis, it also helped to understand the rapid course of the condition, as the mutation was homozygous.

New treatments that act at several levels have been developed to detain or delay transthyretin amyloid deposit.[5] Some have proven to be effective in randomized clinical trials and have been approved by regulatory agencies. Several of these authorized drugs act by inhibiting hepatic expression of transthyretin with interfering ribonucleic acid (patisiran) or antisense oligonucleotides (inotersen). Other drugs act by stabilizing the transthyretin molecule and preventing its dissociation and deposition. This group includes tafamidis, which has been proven to reduce cardiovascular mortality and hospital admissions.[6] Other stabilizers are under development. Finally, it may be possible to eliminate amyloid deposits by antibodies directed toward transthyretin or by molecules such as doxycycline. Several trials are currently underway to evaluate these compounds.[5]

Genetic screening enabled identification of 15 carriers who require close monitoring and may benefit from the early start of these new treatments to slow the development of the disease and improve the prognosis.

An early diagnosis in our patient would have allowed initiation of effective drug treatment and avoided progression to the terminal phase requiring transplantation. Fortunately, the carriers identified, particularly the sister in an early phase of the disease, will benefit from the available treatments.

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REFERENCES

1. García-Pavía P, Tomé-Esteban MT, Rapezzic C. Amiloidosis. También una enfermedad del corazón. *Rev Esp Cardiol.* 2011;64:797–808.
2. Jacobson DR, Pastore RD, Yaghoubian R, et al. Variant-sequence transthyretin (isoleucine 122) in late-onset cardiac amyloidosis in black Americans. *N Engl J Med.* 1997;336:466–473.
3. Ruberg FL, Maurer MS, Judge DP, et al. Prospective evaluation of the morbidity and mortality of wild-type and V122I mutant transthyretin amyloid cardiomyopathy: The transthyretin amyloidosis cardiac study (TRACS). *Am Heart J.* 2012;164:222–228e1.
4. Reddi HV, Jenkins S, Theis J, et al. Homozygosity for the V122I mutation in transthyretin is associated with earlier onset of cardiac amyloidosis in the African American population in the seventh decade of life. *J Mol Diagn.* 2014;16:68–74.
5. González-López E, López-Sainz A, García-Pavía P. Diagnosis and treatment of transthyretin cardiac amyloidosis. Progress and hope. *Rev Esp Cardiol.* 2017;70:991–1004.
6. Maurer MS, Schwartz JH, Gundapaneni B, et al. Tafamadis treatment for patients with transthyretin amyloid cardiomyopathy. *N Engl J Med.* 2018;379:1007–1016.

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Transcatheter Repair of Superior Sinus Venosus Atrial Septal Defect With Partial Anomalous Pulmonary Venous Drainage With the Chimney Double Stent Technique

Reparación percutánea de comunicación interauricular tipo seno venoso superior y drenaje venoso pulmonar anómalo parcial con técnica de doble stent *en chimenea*

To the Editor,

Superior sinus venosus atrial septal defect (SSV-ASD) is an uncommon abnormality of the interatrial septum that is usually accompanied by partial anomalous pulmonary venous drainage (PAPVD) of the right upper pulmonary veins into the superior vena cava (SVC). It is surgically repaired via patch closure of the defect and reconnection of the pulmonary veins to the left atrium (LA). Transcatheter closure has been recently achieved through placement of a coated stent in the SVC.[1] Here, we present the first reported case of transcatheter closure in Spain involving a modified double chimney stent technique.

This case concerns a 58-year-old patient with morbid obesity who exhibited progressive exertional dyspnea. Echocardiography showed right-cavity volume overload. Cardiac magnetic resonance imaging and computed tomography (CT) revealed SSV-ASD with PAPVD with a Qp:Qs of 2.1:1 and normal pulmonary pressure (Figure 1A–C), in addition to a persistent left SVC. Due to the comorbidity, we decided to perform transcatheter closure of the defect. A 3-dimensional cardiac model was printed from the CT images with ITK-SNAP software. The final geometry was exported as an STL file to the Meshmixer program (Autodesk Inc, United States) for modeling. A 0.8-mm outer layer was added and the model was processed by the Cura program (Ultimaker BV, the Netherlands) and sent to a 3-dimensional printer (BQ Witbox, Spain). The model was manufactured using fused deposition modeling technology with polyurethane filament.[2–4]

The procedure was simulated in vitro via implantation of 2 stents overlapping in the SVC and flaring of the proximal stent until the SSV-ASD in the roof of the right atrium (RA) was determined to be sealed. In addition, 2 introducers were inserted from the anomalous pulmonary veins through the ASD to the LA and fluoroscopy was used to verify that there was sufficient space

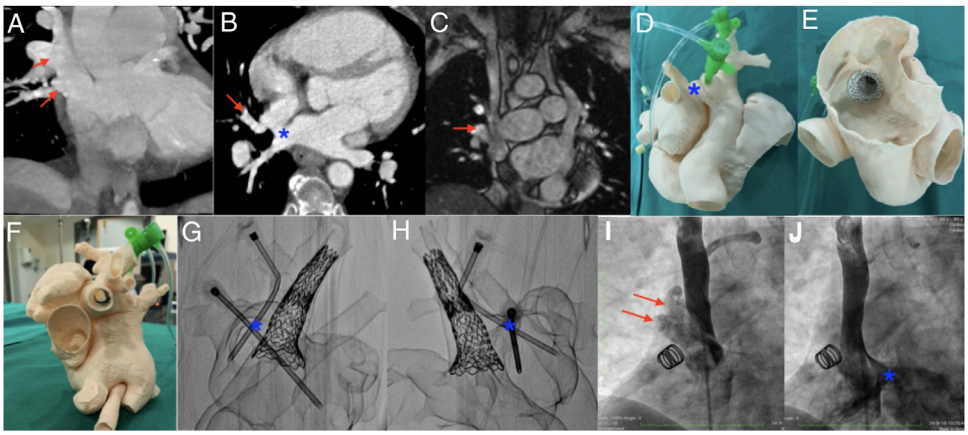

Figure 1. Multimodality in surgical planning. A and B: cardiac computed tomography; the arrows indicate the upper and right middle pulmonary veins draining into the SVC; the asterisk marks the SSV-ASD. C: cardiac magnetic resonance imaging showing the PAPVD (arrow) and ASD. D–F: three-dimensional model; the introducers are observed in the anomalous pulmonary veins, as well as the stent flared from the atrial aspect sealing the defect and the other stent from the SVC; the asterisk marks the SSV-ASD. G and H: X-ray image of the 3-dimensional model showing the stents and the introducers; the asterisk marks the site of the ASD. I and J: angiogram of the SVC showing the anomalous pulmonary veins (arrows) and the SSV-ASD (asterisk). PAPVD, partial anomalous pulmonary venous drainage; SSV-ASD, superior sinus venosus atrial septal defect; SVC, superior vena cava.

between the stents and the wall of the SVC to allow redirection of the flow of the PAPVD to the LA (Figure 1D–H).

Through right femoral and jugular venous and left arterial access, the procedure was initiated by creating a venovenous loop between the internal jugular vein and the femoral vein to facilitate stent implantation and flaring. A 4-Fr Glidecath catheter (Terumo, Europe) was retrogradely introduced through the arterial access until it was located within the PAPVD in order to confirm its patency during the procedure. Baseline angiography was performed to visualize the PAPVD and SSV-ASD (Figure 1I–J and Video 1 of the supplementary material). Via a 16-Fr sheath introduced through the right femoral vein, an 18 × 48 mm coated BeGraft stent (Bentley, Germany) was implanted in the SVC (Figure 2A); this stent was overlapped by another BeGraft stent of 24 × 48 mm. With a 30-mm Z-MED balloon (Numed, United States), the proximal stent was postdilated at the RA while an 18-mm ATLAS balloon (Bard, United States), introduced from the SVC, was simultaneously inflated at the stent overlap site to prevent migration of the proximal stent to the RA during the flare maneuver (Figure 2B). Finally, adequate flow and the absence of a gradient between the PAPVD and the LA were confirmed (Figure 2C-D). Before patient discharge, CT showed the patency of the stents from the SVC to the RA and the redirection of the PAPVD flow between the wall of the SVC and the stents, crossing the anatomical ASD to the LA (Figure 2E–H and Video 2 and Video 3 of the supplementary material).

In summary, planning with the 3-dimensional model allowed exact reproduction of the manipulations required during the actual procedure with the same stents. Through the modeling, a balloon inflation test in the SVC was avoided, although this test is

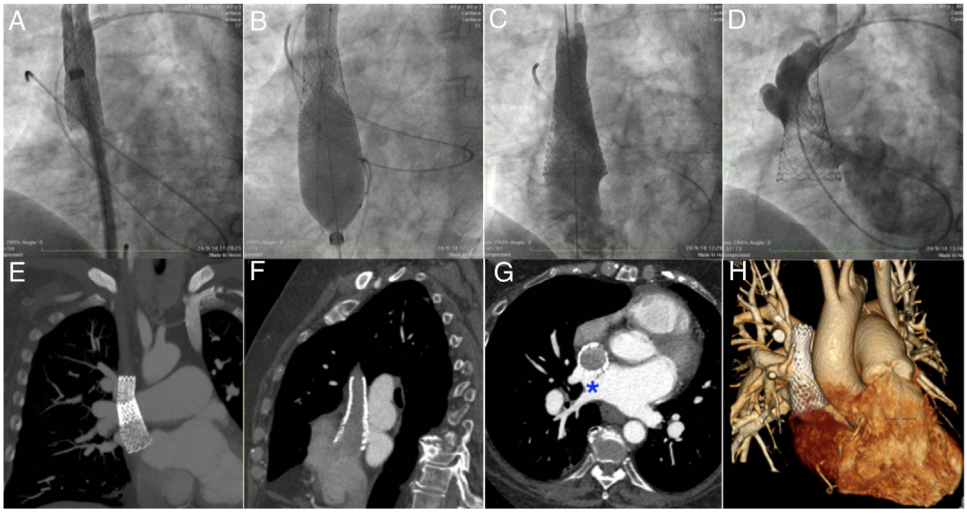

Figure 2. Transcatheter procedure. A: implantation of the more distal stent in the SVC. B: balloon-mediated flaring of the proximal stent while another inflated balloon is maintained in the overlapping zone of the stents to prevent their migration. C: angiogram of the SVC showing complete sealing of the SSV-ASD. D: injection from the retrograde catheter into the PAPVD demonstrating the patency of the anomalous pulmonary veins draining into the LA. E–H: computed tomography images showing the stents, the redirection of the PAPVD flow between the SVC and the stents, and its passage through the anatomical SSV-ASD (asterisk) to the LA. LA, left atrium; PAPVD, partial anomalous pulmonary venous drainage; SSV-ASD, superior sinus venosus atrial septal defect; SVC, superior vena cava.

recommended to verify the absence of PAPVD occlusion.[2,3] Second, for complete sealing of the ASD, the proximal stent must be flared in the roof of the RA and the shortening and maximum diameter of the stent accurately studied. Third, to prevent stent migration during the flare maneuver, the stent should be fixed in place with another balloon. Fourth, if PAPVD occlusion occurs, the catheter can be exchanged for a high-support guidewire and the balloon retrogradely inflated.

Given these technical and planning aspects, and in the absence of more experience, we believe that closure of the SSV-ASD is feasible and safe in patients with contraindication and/or high surgical risk.

CONFLICTS OF INTEREST

Á. Sánchez-Recalde is an Associate Editor of *Revista Española de Cardiología*.

APPENDIX. SUPPLEMENTARY DATA

Supplementary data associated with this article can be found, in the online version, at https://doi.org/10.1016/j.rec.2019.08.004.

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REFERENCES

1. Riahi M, Velasco Forte MN, Byrne N, et al. Early experience of transcatheter correction of superior sinus venosus atrial septal defect with partial anomalous pulmonary venous drainage. *Eurointervention.* 2018;14:868–876.
2. Valverde I. Three-dimensional Printed Cardiac Models: Applications in the Field of Medical Education, Cardiovascular Surgery, and Structural Heart Interventions. *Rev Esp Cardiol.* 2017;70:282–291.
3. Gomez G, Valverde I. 3 D Printing in Revista Española de Cardiología: Just a Passing Fad? *Rev Esp Cardiol.* 2019;72:275.
4. Valverde I, Gomez-Ciriza G, Hussain T, et al. Three-dimensional printed models for surgical planning of complex congenital heart defects: an international multicentre study. *Eur J Cardiothorac Surg.* 2017;52:1139–1148.

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Pre-emptive Left Main Stenting During Transcatheter Aortic Valve Implantation. A Viable Option?

Stent *profiláctico en chimenea en el tronco coronario durante implante percutáneo de válvula aórtica. ¿Una opción adecuada?*

To the Editor,

Surgically implanted prosthesis dysfunction is one of the clinical situations in which percutaneous implantation of a new prosthesis (procedure known as valve-in-valve) has produced good outcomes.[1] One of the major concerns in these cases is the risk of coronary obstruction, which has an incidence of around 2.3% to 3.5%.[1,2] The development of coronary obstruction is related to displacement of the leaflets of the malfunctioning bioprosthesis toward the coronary ostium following expansion of the percutaneous valve.[2] This complication is associated with high mortality and can occur immediately after the procedure or within days (in the VIVID registry, 36.1% of obstructions were delayed).[2,3] There are reports of chimney stent implantation in patients who experience coronary obstruction during prosthesis release.[4,5] However, no publications describe the use of this type of procedure for pre-emptive left main stenting to avoid late obstruction. We report a case with a step-by-step description of this technique.

A 79-year-old man with a history of inferior infarction with papillary muscle rupture in 2003 underwent surgical implantation of a 27-mm Sorin Bicarbon mechanical prosthesis and saphenous vein graft to the anterior interventricular artery. In 2010, a 21-mm Mitroflow biological prosthesis was implanted due to severe degenerative aortic stenosis.

In September 2018, the patient was admitted for heart failure secondary to aortic bioprosthesis dysfunction, with severe intraprosthetic aortic regurgitation. Echocardiography also showed severe left ventricular dysfunction, with inferoposterior necrosis, and a normal-functioning mitral mechanical prosthesis. The patient had no clinical, laboratory, echocardiographic, or microbiologic evidence of infective endocarditis, which was definitively ruled out by positron-emission tomography. Coronary angiography showed severe 3-vessel coronary disease with a patent saphenous vein bypass to the anterior interventricular artery (figure 1AA). To evaluate the valve complex and vascular accesses, computed tomography angiography was performed, detecting as the greatest limitation for percutaneous aortic valve implant a low height of the left main coronary artery (LMCA) (height of 8.6 mm above the prosthetic ring) (figure 1B) and narrow sinuses of Valsalva. The distance from the aortic prosthetic ring to the mitral prosthesis was 9.9 mm (figure 1C). In the simulated reconstruction, the distance at the sinuses of Valsalva between the virtual transcatheter ring and the origin of the LMCA was 3.3 mm (figure 1D). A distance < 4 mm has been associated with a high risk of coronary obstruction.[2]

The patient was readmitted in December 2018 due to a new episode of refractory heart failure. The case was discussed in the medical-surgical session and, in view of the high surgical risk (logistic EuroSCORE of 47.2% and Society of Thoracic Surgeons score of 11.7%), a decision was made to implant an Evolut R^{TM} valve (Medtronic; Minneapolis, Minnesota, United States) of 23 mm.

The procedure via femoral access was planned for pre-emptive chimney stenting in the LMCA due to the high risk of coronary obstruction, in light of the external position of the Mitroflow valve leaflets, the measurements reported, and recent reports that a third of all obstructions are delayed.[2] The procedure was performed under general anesthesia and transesophageal ultrasound guidance. Double femoral and double radial access was used to protect