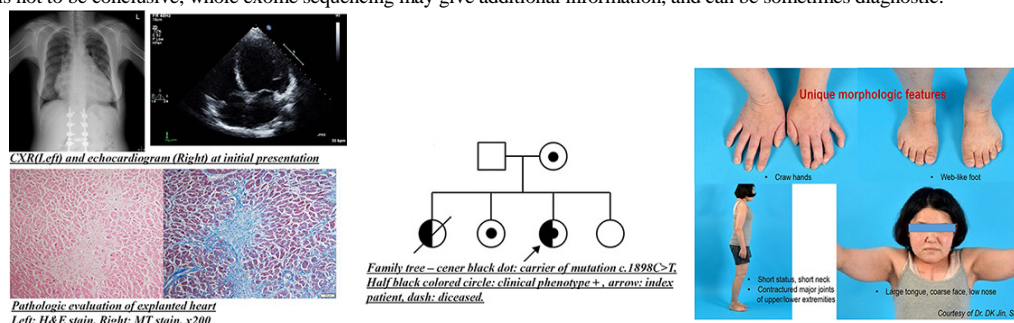


Familial DCMP diagnosed with Mucopolipidosis III Alpha/Beta by Whole Exome Sequencing

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Introduction: Familial DCMP is sometimes combined with systemic genetic syndromes. Among them, patients who have skeletal dysplasia, appropriate diagnostic examination should be performed. **Case Report:** A 33-year-old woman was presented with cardiac arrest from ventricular fibrillation. She had been diagnosed with DCMP 2 years ago. Although she has given medication and ICD several months after resuscitation, her cardiac function remained decreased with left ventricular ejection fraction of 33% on echocardiogram with symptom of NYHA III. She got heart transplantation soon. Pathologic examination of explanted heart showed dilated left ventricle with fibrosis and inflammatory change, but no coronary and valvular abnormalities. We noticed her morphologic features with coarse face, low nose, large tongue, claw hands, contracted major joints in upper and lower extremities. She was 153cm and 56kg. She had spinal surgery from unknown lumbar disease 8years ago. Her sister also diagnosed with DCMP and spinal disease at the age of 35, and died of sudden cardiac death. Under the suspicion of mucopolysaccharidosis(MPS) I, diagnostic tests were done. Urine glycosaminoglycans and serum α -L-iduronidase was marginally diagnostic. Direct sequencing showed c.1898C > T(p.Ser633Leu) heterozygote mutation. Such heterozygote mutation was discovered in a patient with MPS I, we started enzyme replacement therapy for several months. Her mother and sister functionally and morphologically normal but known to have same mutation. Therefore, we performed whole exome sequencing to diagnose genetic mutations with familial DCMP and skeletal dysplasia. Heterozygote mutation in GNPTAB gene with c.2715+1G > A and c.3173C > G was found. Mucopolipidosis III alpha/beta was finally diagnosed after confirmation of elevated plasma enzyme activity of arylsulfatase A and hexosaminidase, and not elevated plasma activity of β -glucosidase. **Discussion:** This is the case of familial DCMP in a 32-year-old woman diagnosed with mucopolipidosis III alpha/Beta by Whole Exome Sequencing. If several test reveals not to be conclusive, whole exome sequencing may give additional information, and can be sometimes diagnostic.



Hemodynamic assessment in patient with mechanical valves using pressure wire

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Invasive catheter-based pressure measurements are technically challenging particularly in cases with mechanical valve. The standard catheter technique for assessing pressure across prosthetic valve can result in fatal hemodynamic collapse from prosthetic leaflet malfunction due to the large catheter size. We report a case that underwent cardiac catheterization using a pressure coronary guide wire to assess trans-pulmonary pressure gradient to investigate the feasibility for heart transplantation. A 62-year-old woman admitted with gradual worsen dyspnea. She previously had cardiac surgery three times, latest surgery was mitral valve, aortic valve and tricuspid valve replacement surgery at 2008. On the echocardiography, there were dehiscences and paravalvular leakage at posteromedial site and lateral site of prosthetic mitral valve. Physiologic severe mitral regurgitation was observed. The patient was considered heart transplantation, by reason of reduced left ventricular ejection fraction, and technical difficulty because of her 4th re-do mitral valve operation. Invasive catheterization was performed to evaluate the presents of pulmonary arterial hypertension, the relative contra-indication of heart transplantation. Right heart catheterization was performed using a 4-French glide catheter, across the prosthetic tricuspid valve. However, pulmonary wedge pressure could not be obtained due to difficulty of catheter advance into the segmental level of pulmonary artery. Left hemodynamic assessment was performed using 0.014-inch coronary Pressure Wire across the mechanical aortic and mitral prosthesis through a 4 French glide catheter. Transpulmonary gradient was 27 mmHg. Cardiac Index was 2.47 L/min/m². Pulmonary vascular resistance was 8.0 Wood units. After catheterization, there was no fluoroscopic evidence of a change in valve function. Owing to the high pulmonary vascular resistance, the patient did not take heart transplantation. The patient underwent a successful fourth sternotomy with mitral and aortic valve replacement and aorto-mitral fibrous body reconstruction with bovine pericardium.

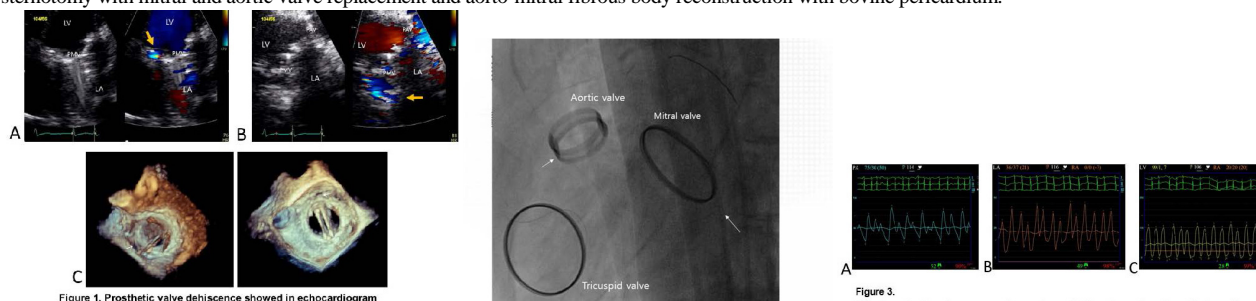


Figure 1. Prosthetic valve dehiscence shown in echocardiogram (A) Prosthetic valve dehiscence showed at posteromedial site of mitral valve in apical 4 chamber view of transthoracic echocardiogram. (B) Also, dehiscence showed at lateral site of mitral valve in parasternal long axis view. (C) 3D reconstruct image in trans-esophageal echocardiogram. Dehiscence site marked with white arrow.

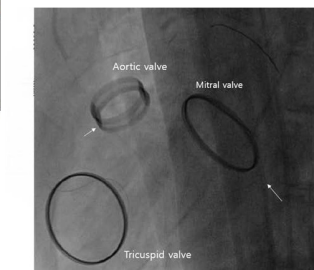


Figure 2. Invasive cardiac catheterization. Fluoroscopic image showing the 0.014-inch high-fidelity coronary pressure wire pass through aortic valve (short arrow) and mitral valve (long arrow).

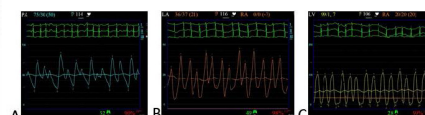


Figure 3. Hemodynamics from the pressure wire was transmitted to a hemodynamic monitoring system. (A) FA wave (B) LA wave (C) LV wave.