# Polymorphic Ventricular Tachycardia And Complete Heart Block In Hunter Syndrome

L Shavit, M Joachim, R Nir-Paz, T Grenader

**Citation**

L Shavit, M Joachim, R Nir-Paz, T Grenader. *Polymorphic Ventricular Tachycardia And Complete Heart Block In Hunter Syndrome*. The Internet Journal of Internal Medicine. 2005 Volume 6 Number 1.

**Abstract**

We present a 27-year-old male patient with Hunter syndrome (mucopolysaccharidosis type II) who had severe cardiopulmonary disease including valvular involvement and pulmonary hypertension. Our patient developed polymorphic ventricular tachycardia following by complete atrioventricular block without any obvious trigger. We suggest that the high incidence of unexpected sudden death in patients with mucopolysaccharidosis may be due to spontaneous cardiac arrhythmia.

## INTRODUCTION

Mucopolysaccharidoses is a group of hereditary diseases characterized by a deficiency of one of lysosomal enzymes involved in mucopolysaccharide metabolism. It is usually transmitted as autosomal recessive or sex-linked recessive trait. Cardiac involvement is present in 72-100% of patients with MPS and is especially common and severe in Hurler, Hunter and Maroteaux-Lamy syndromes [1,2,3]. The valves are the most often affected structure regardless of mucopolysaccharidosis type, though endomyocardial, myocardial, coronary artery and conduction system involvement may also occur. In most cases the cause of death is cardiorespiratory failure secondary to cardiovascular disease and/or upper airway obstruction with or without infection. The incidence of unexpected sudden death, most presumed due to cardiac arrhythmia, is approximately 10%.

We report a patient with Hunter syndrome (mucopolysaccharidosis type II), who developed polymorphic ventricular tachycardia and complete atrioventricular block.

## CASE PRESENTATION

A 27-years old man patient was brought to the emergency room with a 3 hours history of sudden onset severe dyspnea and cyanosis.

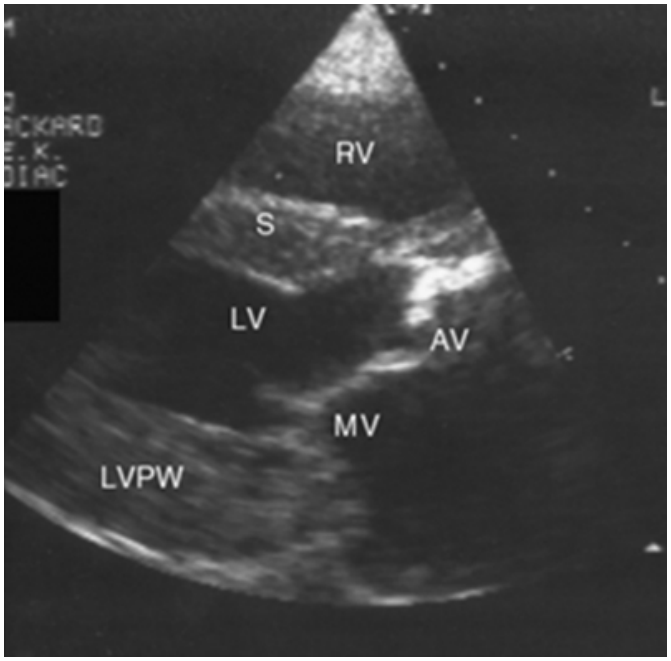
At the age of 2-years he was diagnosed as having Hunter syndrome with typical habitus, retarded speech, deafness and hepatosplenomegaly. His maternal uncle also suffered from Hunter syndrome. Later on diagnosis was confirmed genetically.

Subsequently the patient demonstrated normal mental development, but suffered from recurrent respiratory tract infection due to upper airways obstruction by granulation tissue finally necessitating permanent tracheostomy at the age of eight. The patient continued to suffer from pulmonary infections and recurrent bronchoscopies with excisions of tracheal wall granulations were performed.

At the age of 20 years, cardiac involvement was diagnosed. Echocardiography showed concentric hypertrophy and widespread valvular disease, with mixed mitral valve disease (thickened stenotic mitral valve with severe mitral incompetence and moderate mitral stenosis), and severe aortic stenosis (heavily calcified aortic valve with aortic valve area 0.5cm2 and peak gradient 130 mm Hg). Moderate tricuspid incompetence and moderate pulmonary hypertension (tricuspid valve gradient 56mm Hg) was also demonstrated (Figure 1). Amiloride and hydrochlorthiazide treatment was initiated. Later amiloride was replaced with furosemide and spironolactone.

**Figure 1**

Figure 1: Parasternal long axis view from a two-dimensional echocardiography shows concentric hypertrophy of left (LV) and right ventricle (RV), markedly thickened intraventricular septum (S) and left ventricular posterior wall (LVPW), thickened mitral (MV) and calcified aortic valve (AV).

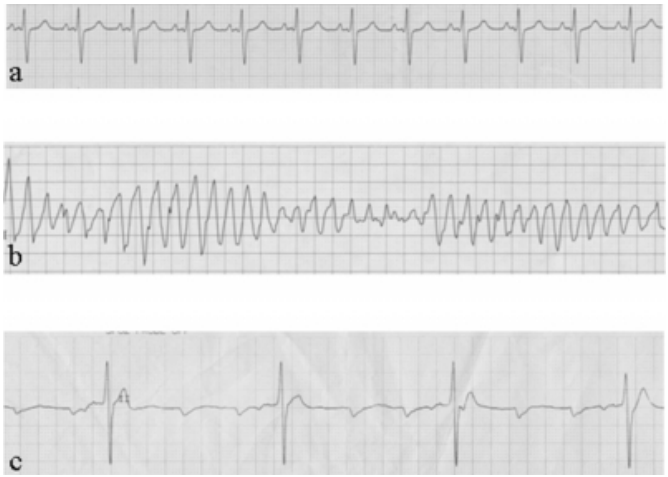



On admission the physical examination revealed the characteristic morphological features of Hunter syndrome: short stature, short neck, coarse face, macroglossia, thickened lips, bridged nose and short thick fingers. The patient was alert and dyspneic. The blood pressure was 130/80mm Hg, pulse rate was 106 beats per minute, and the respiratory rate was 28 breaths per minute. He was afebrile. Jugular venous pressure was maximally increased. Loud rales were heard over both lung fields. Heart sounds were regular and a prominent 3/6 systolic murmur was heard over all precordium. The abdomen was soft and protuberant with marked hepatosplenomegaly.

Laboratory blood tests demonstrated normal complete blood cell count and biochemistry panel. Chest X-ray showed marked cardiomegaly with signs of pulmonary edema. On electrocardiogram there was normal sinus rhythm 96 per minute, left axis deviation, normal PR interval and no QT-prolongation. (Figure 2a)

**Figure 2**

Figure 2: The rhythm strip shows a) normal sinus rhythm with normal QT interval on admission b) polymorphic ventricular tachycardia and c) complete atrioventricular block.




Upper airway obstruction was excluded by bronchoscopy and mechanical ventilation was initiated via tracheostomy. The patient was transferred to the intensive cardiac care unit, and treated with intravenous furosemid, oral spironolactone and broad spectrum intravenous antibiotics.

On the second hospitalization day patient suddenly developed polymorphic ventricular tachycardia (Figure 2b) and sinus rhythm was immediately restored by electrical cardioversion. A few minutes later, the patient became very agitated, dyspnoeic and electrocardiographic monitoring revealed complete atrioventricular block (Figure 2c). Cardiopulmonary resuscitation was undertaken and sinus rhythm restored, but the patient did not regain consciousness. Brain computer tomography revealed chronic hydrocephalus and electroencephalography showed minimal brain electrical activity consistent with anoxic brain damage. The patient died 3 weeks later from sepsis.

## DISCUSSION

We present a patient with Hunter syndrome who developed polymorphic ventricular tachycardia and complete atrioventricular block without any evident triggering factor. Our patient suffered from severe cardiopulmonary disease, including upper airway obstruction, severe valvular involvement with mitral valve thickening, incompetence and stenosis, severe aortic valve stenosis and pulmonary hypertension.

There was no previous electrocardiographic evidence of conduction abnormalities or ischaemic heart disease. There

was no history of chest pain, but coronary artery disease was not excluded. PR and QT intervals were normal. Blood tests did not show any electrolyte abnormalities, and respiratory acidosis and hypoxia were fully corrected by mechanical ventilation at the time of admission.

Cardiac involvement is a frequent and often severe complication of Hunter syndrome. Valvular thickening with regurgitation or stenosis, hypertrophic cardiomyopathy, endocardial thickening, coronary artery stenosis, and pulmonary hypertension are commonly seen. The valvular damage and functional impairment are generally progressive [1,2,3].

Involvement of the cardiac conduction system has been described, and may be secondary to a direct effect of infiltration of conduction tissue by so-called gargoyle cells, large fibroblasts expanded by large amounts of mucopolysaccharides or ischemic damage [4].

We have found 2 case reports describing complete atrioventricular block in patients with mucopolysaccharidoses One case describes complete heart block was triggered by guide wire insertion to the superior vena cava during anesthetic management [5], the other describes spontaneous complete atrioventricular block [6].

To the best of our knowledge there is no report describing polymorphic ventricular tachycardia in patients with mucopolysaccharidoses. In our case we could not demonstrated any obvious trigger for occurrence of cardiac arrhythmia. We suggest that the high incidence of unexpected sudden death in patients with mucopolysaccharidoses may be due to spontaneous malignant cardiac arrhythmia. Consequently a 24 hour Holter electrocardiogram examination and implantable defibrillator may be indicated for all patients with Hunter syndrome and proven cardiac disease.

## References

1. Nelson J, Shields MD, Mulholland HC. Cardiovascular studies in the mucopolysaccharidoses. J. Med. Genet. 1990; 27: 94-100.
2. Dangel JH. Cardiovascular changes in children with mucopolysaccharide storage diseases and related disorders--clinical and echocardiographic findings in 64 patients. Eur. J. Pediatr. 1998; 157: 534-538.
3. Rigante D, Segni G. Cardiac structural involvement in mucopolysaccharidoses. Cardiology. 2002;98:18-20.
4. Okada R, Rosenthal IM, Scaravelli G, Lev M. A histopathologic study of the heart in gargoylism. Arch. Pathol. 1967; 84: 20-30.
5. Toda Y, Takeuchi M, Morita K, et al. Complete heart block during anesthetic management in a patient with mucopolysaccharidosis type VII. Anesthesiology. 2001; 95: 1035-1037.
6. Hishitani T, Wakita S, Isoda T, et al. Sudden death in Hunter syndrome caused by complete atrioventricular block. J. Pediatr. 2000; 136: 268-269.

## Author Information

**Linda Shavit, M.D.**
Department of Medicine, Sha'ree Zedek Medical Center

**Michael Joachim**
Faculty of Medicine, Hebrew University Hadassah

**Ran Nir-Paz, M.D.**
Department of Medicine, Hadassah University Hospital

**Tal Grenader, M.D.**
Department of Oncology, Hadassah University Hospital