



# A Rare Case of Watson Syndrome

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## Abstract

Watson for the first time reported a case series of children in a family that presented with pulmonary valve stenosis, mental retardation, short stature, and small brown color skin lesions that are known as cafe-au-lait spots. We present a rare new variant of the syndrome in an adult patient with severe pulmonary valve stenosis, main, left, and right pulmonary artery aneurysm, short stature, mental retardation, coronary artery disease, and skin lesions. The patient underwent open cardiac surgery with pulmonary valvotomy and aneurysmorrhaphy of the main pulmonary artery and its right and left branches. The postoperative course was uneventful and the six-month follow-up with transthoracic echocardiography revealed no recurrence of aneurysm of repairing pulmonary arteries and good clinical outcome of the patient. Our patient had a unique characteristic of aneurysm of the main pulmonary artery and its both branches that has rarely been reported previously in the medical literature.

## Keywords

adult, congenital heart disease, neurofibromatosis, Watson syndrome

## INTRODUCTION

Genetic disorders are not uncommon in congenital heart defects and a wide variety of genetic related anomalies have been reported in the medical literature. Association of skin and cardiac anomalies that is defined as the cardio-cutaneous syndrome has been reported in a variety of syndromes and one of the rarest types of these is the Watson syndrome. Other common types of this association include Noonan and LEOPARD syndrome.<sup>[1]</sup> Since 1967, when the syndrome was described in the literature, the associated anomalies of the syndrome have been widened to include neurofibromatosis, frontal macrocephaly, and skin lesion in 30% of affected subjects.<sup>[2,3]</sup> Major differences in regards to the incidence of syndrome components have been reported in each case in the medical literature. In some cases, patients present with cardiac defect and neurofibromatosis without specific skin lesions and the others were diagnosed with cardiac defect, Lisch nodules and skin spots.<sup>[4]</sup>

Molecular genetics examination demonstrated the presence of several adjacent genes for the appearance of pulmonary stenosis, skin anomalies, short stature, and mental retardation on deletion in the long arm of the chromosome.<sup>[5]</sup> We report here an extremely rare case of Watson syndrome in a 45-year-old man that was successfully treated surgically.

## CASE REPORT

A 45-year-old man was admitted to the hospital with unstable chest pain. His medical history revealed the presence of some degree of mental retardation, lower extremities, and skin spots, but evidence of characteristic lesions of neurofibromatosis was not found. Physical examination showed a short stature man with mental retardation and lower extremity skin spot lesions.

Cardiac auscultation exhibited that a systolic murmur was maximum detected in the left third intercostal space.

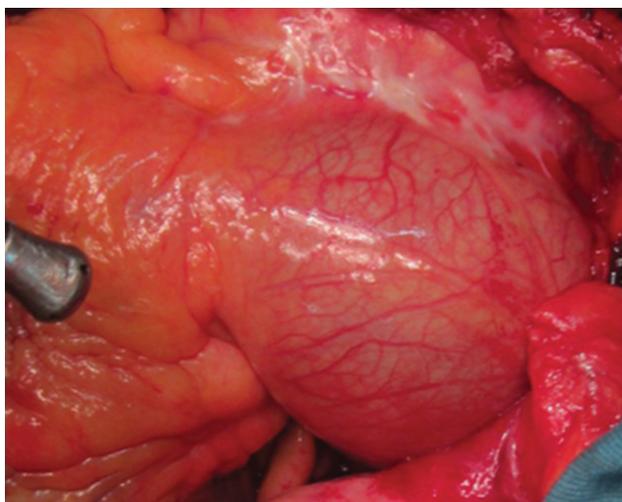
A bulging on the pulmonary trunk area on the chest x-ray (Fig. 1) prompted us to perform a transthoracic (TTE) and transesophageal echocardiography (TEE) (Fig. 2). TEE revealed a 4.27 cm size of the main trunk and 3.4 cm size of the right pulmonary artery aneurysm. The left pulmonary artery measured 3.2 cm in TTE. TTE revealed dilated right ventricular size with reduced function and severe valvular and subvalvular pulmonary artery stenosis. Cardiac angiography showed severe left anterior descending coronary artery stenosis. No other congenital anomalies of the coronary artery were detected and left ventricular function was preserved. The patient was scheduled for open cardiac surgery with aortic and bi-cava cannulation. Upon pericardiotomy, palpation of the pulmonary valve area revealed a thrill of harsh murmur with aneurysmal dilatation of the main, left, and right pulmonary arteries. The aneurysmal dilatation of pulmonary arteries, pulmonary valve, and subvalvular stenosis was confirmed intraoperatively by TEE (70 mm Hg gradient). The pulmonary artery pressure values were normal as measured by the right ventricle to the pulmonary artery gradient. The pulmonary valve with its typical congenital commissural fusion was treated by valvuloplasty that consists of triple commissurotomy and evaluation of subvalvular obstructive lesion. Subvalvular outflow tract obstruction was managed by infundibular myotomy and resection of some obstructing muscle bundle. The main pulmonary artery and its left and right branches were aneurysmally dilated immediately above the stenotic pulmonary valve (Figs 3, 4). After dissection of the main pulmonary artery and its left and right branches from surrounding tissue, the dissection continued to the pulmonary hilar part of the right and left pulmonary artery branches where the lobar branches were separated and entered the lung tissue. A longitudinal strip of the anterior wall of the main pulmonary trunk (2-3 cm) and left and right pulmonary arteries (1.5 cm) was resected. Aneurysmorrhaphy with continuous suture line (proline 4/0) continued to



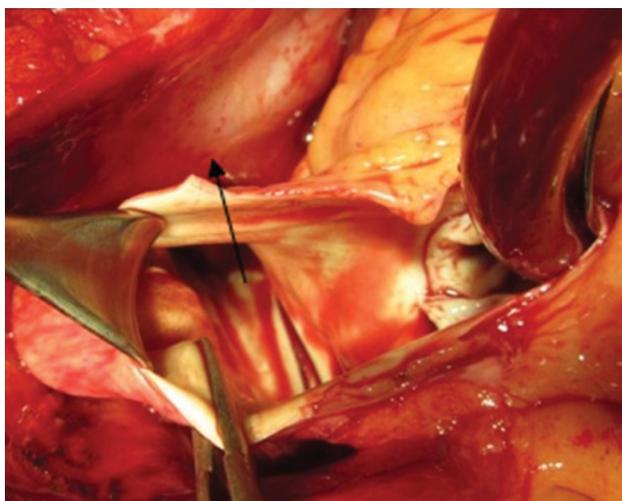
**Figure 1.** Chest x-ray shows lobar pulmonary arteries dilatation in both lung field.



**Figure 2.** Preoperative TEE shows dilated main (4.27 cm) and the right and left (3.2 cm) pulmonary arteries aneurysm. TTE revealed dilated right ventricular size and severe valvular and subvalvular pulmonary artery stenosis.

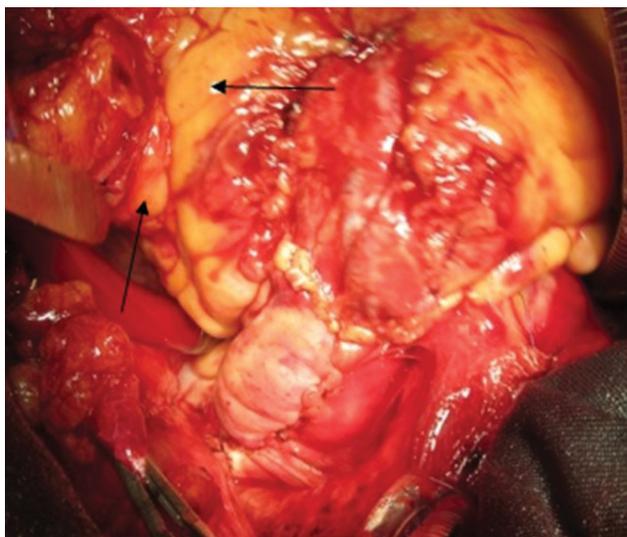


**Figure 3.** Dilatation of the main pulmonary artery and its left and right branches above the stenotic pulmonary valve.



**Figure 4.** Dilated main, left, and right pulmonary artery above the stenotic pulmonary valve (black vertical arrow).

the pulmonary valve ring, and then a fresh pericardial patch was used for the right ventricular outflow tract (RVOT) patch angioplasty including the pulmonary valvular ring (Fig. 5). A single left anterior descending coronary artery (LAD) bypass was then performed by anastomosis of the left internal mammary artery (LIMA) to the LAD. After weaning from cardiopulmonary bypass (CPB) with the good hemodynamic condition, pulmonary valvar gradient reduced to 5 mm Hg, with mild pulmonary regurgitation on postoperative TEE. Postoperative TEE also revealed a proper reduction in size of the main and left pulmonary artery; however, the recorded size of the distal portion of the right pulmonary artery was 2.6 cm (preoperative size: 3.4 cm) (Fig. 6). Few hours



**Figure 5.** Repaired main pulmonary artery (vertical black arrow). Aneurysmorrhaphy with continuous suture line (proline 4/0) continued to pulmonary valve ring. Patch angioplasty of right ventricular outflow tract (transverse black arrow).



**Figure 6.** Postoperative size of the right pulmonary artery (B: 2.19 cm) and main pulmonary artery (A: 2.62 cm). Shadow in the main pulmonary artery caused by postoperative pulmonary regurgitation.

after patient's arrival to the intensive care unit and cessation of chest tube drainage, heparin was initiated at 1000 U per hour and warfarin was prescribed orally via nasogastric tube (5 mg). When prothrombin time was raised to 18-20 seconds, we stopped heparin and treatment continued with acetylsalicylic acid (81 mg/day). The postoperative course was uneventful, and the patient was discharged home 10 days postoperatively. Histopathologic evaluation of the pulmonary artery wall revealed no evidence of the other rare connective tissue disorder.

## DISCUSSION

Our patient presented with a rare complex of coronary artery disease (CAD), the main and both pulmonary branches aneurysm, congenital pulmonary valve stenosis, mental retardation, and skin brown spot lesion. Our surgical approach includes bypassing his LAD stenosis with a LIMA graft. The patient's pulmonary stenosis was amenable to repair by commissurotomy and it was repaired with this mechanism. However, in cases with severely calcified or fibrotic valve, valve replacement could be considered with a biologic or homograft mechanism. Watson syndrome, with this complex pathological evidence, is a rare event, and there are no large case studies in the literature. Due to the paucity of reported cases, the natural history of medically treated subjects is unknown, although some exceptional cases of lethal dissection of the pulmonary wall or rupture to surrounding structures have been reported.<sup>[6]</sup> The accepted surgical strategy has included an aneurysmorrhaphy with resection of a strip of anterior wall of aneurysm or replacement of the main trunk with a prosthetic Dacron graft or homograft.<sup>[7]</sup> The aneurysm in our case involved not only the main pulmonary trunk but also both the right and left pulmonary arteries. This pathologic finding is easily amenable to repair by longitudinal aneurysmorrhaphy through a median sternotomy. Post-stenotic dilatation was the most probable cause of the aneurysm in our case, with the absence of pathologic finding of the basic connective tissue disorder.

The Watson syndrome is an autosomal dominant condition that is allelic to type 1 neurofibromatosis (NF1). It has been reported that pulmonary stenosis is contiguous to NF1 gene. Although they have the same manifestations, pulmonary stenosis is more common in Watson syndrome. Our patient fit the criteria for the Watson syndrome with detection of clinical findings such as mental retardation, short stature, skin spots, pulmonary valve stenosis and aneurysm, and CAD. However, due to study limitations, we were unable to examine the patient's genetic structure. DNA studies will be required for further investigation.

Our case report is probably the second description of the surgical approach to pulmonary stenosis, pulmonary artery aneurysm, and coronary disease with this syndrome.

## CONCLUSIONS

We present a rare, new variant of the Watson syndrome in an adult patient with severe pulmonary valve stenosis, main, left and right pulmonary artery aneurysm, short stature, mental retardation, coronary artery disease and skin spots lesions. Our patient has a unique characteristic of aneurysm that has rarely been reported previously in the medical literature.

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### Резюме

Ватсон впервые сообщил о серии случаев у детей в семье со стенозом клапана лёгочной артерии, умственной отсталостью, низким ростом и небольшими поражениями кожи коричневого цвета, известными как пятна типа „кофе с молоком“. Мы представляем редкий новый вариант синдрома у пожилого пациента с тяжёлым стенозом клапана лёгочной артерии, аневризмой главной, левой и правой лёгочных артерий, низким ростом, умственной отсталостью, ишемической болезнью сердца и поражением кожи. Больному выполнена открытая операция на сердце с вальвотомией лёгкого и аневризморфией магистральной лёгочной артерии и её правой и левой ветвей. Послеоперационный период протекал без осложнений, при динамическом наблюдении в течение полугода с помощью трансторакальной эхокардиографии рецидива аневризмы восстанавливаемых лёгочных артерий не выявлено, а клинический исход у больного хороший. У нашего пациента была уникальная характеристика аневризмы главной лёгочной артерии и обеих её ветвей, о которой ранее редко сообщалось в медицинской литературе.

### Ключевые слова

пожилой, врождённый порок сердца, нейрофиброматоз, синдром Ватсона