

Truncus arteriosus communis with survival to the age of 46 years: case report

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SUMMARY

Truncus arteriosus communis is an uncommon congenital cardiovascular malformation characterized by a single arterial trunk that arises from the base of the heart and gives rise to the coronary, pulmonary and systemic arteries. The prognosis in truncus arteriosus is very poor without surgical correction. The median age at death without surgery ranges from 2 weeks to 3 months, with 85 % mortality by age 1 year. The authors report the autopsy findings of a 46 year old man with truncus arteriosus communis without surgical intervention who died at the hospital shortly after admission.

Keywords: truncus arteriosus communis – heart failure – autopsy findings

Truncus arteriosus communis s prežívaním 46 rokov: kazuistika

SÚHRN

Súdny lekár sa častokrát stretáva s prípadmi náhleho a neočakávaného úmrtia v zdravotníckom zariadení, u ktorých nie sú známe anamnestické údaje o ochoreníach, prípadne tieto údaje sú nedostatočné. Pitva často odhalí nálezy, ktoré sú neobvyklé. Medzi takéto raritné nálezy patria aj rôzne vývinové chyby kardiovaskulárneho systému. Autori prezentujú pitevný nález u 46-ročného muža s truncus arteriosus communis bez chirurgickej intervencie, ktorý zomrel krátko po prevoze do nemocnice za známok kardiorespiračného zlyhania. Truncus arteriosus communis je zriedkavá vrodená kardiovaskulárna anomália, pri ktorej výtoková časť pravej aj ľavej komory ústi priamo do spoločného arteriálneho kmeňa s jednou súpravou chlopní, ktorý obstaráva koronárnu, pľúcnu a systémovú cirkuláciu. Táto zriedkavá anomália je výsledkom zlyhania septácie primitívneho arteriálneho trunku počas embryonálneho života. Jednotlivé typy sú definované na základe miesta odstupe pľúcnych tepien z arteriálneho kmeňa. Celosvetovo predstavuje asi 1-2 % všetkých vrodených vývinových chýb srdca. Incidencia je 5-15 prípadov na 100 000 živonarodených detí. Ide o malformáciu, ktorá bez chirurgickej liečby má veľmi zlú prognózu. Bez liečby je táto kardiovaskulárna anomália zvyčajne fatálna a len asi 15 % jedincov prežije 1 rok života. Celkom výnimočne sa môžu títo jedinci dožiť vyššieho veku aj bez chirurgickej intervencie ako to bolo aj v prezentovanom prípade. Operačné riešenie truncus arteriosus communis nebolo vykonané pre technickú nedostupnosť na Slovensku v čase diagnostikovania anomálie u tohto muža.

Kľúčové slová: truncus arteriosus communis – srdcové zlyhanie – pitevný nález

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Forensic pathologists often investigate deaths that occur suddenly and unexpectedly shortly after hospital admission. Obtaining information may be limited at this time, especially when the deceased medical history is unavailable. Autopsy may reveal unusual findings, such as rare cardiovascular malformations. Therefore, choosing proper dissection techniques to clarify the cause of death is essential.

Truncus arteriosus communis is an uncommon congenital cardiovascular anomaly characterized by a single arterial trunk that arises from the base of the heart and gives rise to the coronary, pulmonary and systemic arteries. A single semilunar valve is found in truncus arteriosus. The anomaly is thought to result from incomplete or failed septation of the embryonic truncus arteriosus. Truncus arteriosus represents 1-2 % of congenital he-

art defects in liveborn infants (1,2). It occurs in approximately 5-15 of 100 000 live births (2). The authors present the case of a man with truncus arteriosus communis with survival to the age of 46 years without any surgical correction.

CASE REPORT

We report the case of a 46 years old man with mild intellectual disability and congenital cardiovascular malformation – truncus arteriosus communis. After the autopsy, we requested to view his complete medical records and learned that he had nine siblings. Two of his brothers died of an unspecified congenital heart disease. Initially he was diagnosed with tetralogy of Fallot in 1972 when he was 6 years old. In 1975 this diagnosis was modified to truncus arteriosus communis and the condition was deemed inoperable at that time. He overcame common childhood illnesses and was repeatedly hospitalized for bronchopneumonia and cystitis as a child.

He was treated for arterial hypertension and in 2010 had episodes of grand mal seizures with cerebral hypoxia. From July till December 2011 he was hospitalized four times at the Department of Pneumology and Phtiseology and once at the Department of Anesthesiology and Intensive Medicine. The reason for his admission was recidiving hemoptysis and hemoptoe.

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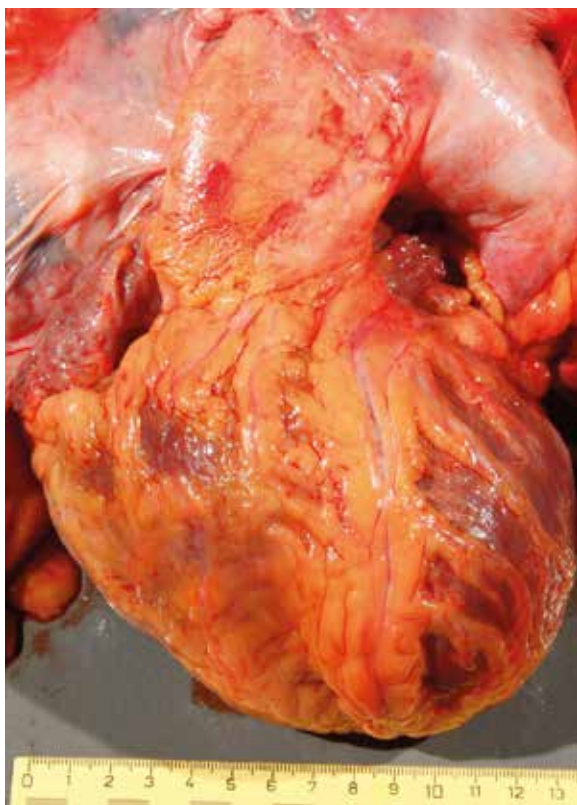


Fig. 1. The heart from the front showing a single large arterial vessel arising from the base of the heart.

The CT scan revealed extensive intraparenchymal pulmonary hemorrhage in the middle and lower lobe of the right lung with the source of bleeding in the S7 segment. At that time surgical intervention was contraindicated for the patient's condition and anatomical localization of the bleeding. He received conservative treatment and was given hemostyptic drugs. Also he was diagnosed with secondary polyglobulia following blood tests. His last admission to the hospital was in May 2012 for hemoptysis, dyspnea and acute respiratory failure that required immediate resuscitation. He died 50 minutes after admission. According



Fig. 2. Opened right and left ventricle of the heart. A single large artery which contains three semilunar valve cusps arises from the summit of the two ventricles.

to the patient's clinical data the immediate cause of death was heart failure due to unspecified congenital malformation of the heart. Autopsy was requested by the medical staff.

At autopsy the body was that of a male adult who appeared about the stated age of 46 years. The body was normally built and well-nourished. Skin was essentially unremarkable except for the presence of abnormal blue discoloration of the skin and mucous membranes (cyanosis). Clubbing of the fingers and toes was evident. The significantly enlarged heart weighted 656 g. There was considerable hypertrophy of both ventricles. The thickness of the right ventricular wall was 0,9 cm and the left ventricular wall thickness was 2 cm. A single large arterial vessel arose from the base of the heart (Fig. 1). The proximal part of the ventricular septum was not formed. The truncal valve was tricuspid – it contained three large semilunar cusps (Fig. 2). The circumference of the arterial trunk 1 cm above the truncal valve was 9 cm. The pulmonary arteries arose from the left posterolateral aspect of the truncus arteriosus, the right pulmonary artery 8,5 cm above the truncal valve and the left pulmonary artery 9 cm above the truncal valve. The aortopulmonary septum wasn't developed. The arterial trunk transitioned to aortic arch giving off its three major branches. The arterial trunk and the coronary arteries showed moderate atherosclerotic changes. There was an isolated old infarction on the cut surface of the right ventricle 2,5 x 1,5 cm in size. Severe dispersed myofibrosis on the cut surface of the anterior and posterior wall of the left ventricle was macroscopically evident. Pulmonary edema and numerous confluent pleural hemorrhages were present. The right lung weighed 670 grams and the left lung 520 grams. The liver appeared normal in size and the cut surface didn't show nutmeg pattern. The left kidney demonstrated hydronephrosis with hydroureter without any source of obstruction. There were no macroscopically evident pathomorphological changes in other organs.

Microscopically well-healed myocardial infarct of the right ventricle with dense collagenous scar was evident. The anterior and posterior wall of the left ventricle showed severe dispersed myofibrosis with significant sclerotic changes of the intramural coronary arteries. There was no sign of chronic passive congestion of the liver. Pulmonary hemorrhage was present. Lung section specimen stained with Perl's Prussian blue showed a large number of hemosiderin-laden macrophages (siderophages).

DISCUSSION

Truncus arteriosus communis is an uncommon congenital cardiovascular malformation. Although there are several case reports of patients with truncus arteriosus surviving to middle age without surgery, the natural history of this condition usually runs a much shorter course (3). In one autopsy series the median age of survival of 94 patients with the disease reported up to 1962, was only five weeks (4). Another series reported a survival of only 15 % beyond the age of one year (2,5,6). In 1935 Carr, Goodale and Rockwell reported the case of a man with truncus arteriosus communis who lived to the age of 36 years (7). In 1966 Hicken, Evans and Heath presented the case of a 38 years old woman with this anomaly (8). Probably the longest survival reported is the case of a patient with truncus arteriosus with survival to the age of 52 years (9).

In 1949 Collett and Edwards recognized four types of truncus arteriosus on the basis of the anatomic origin of the pulmonary arteries. In type I, a short pulmonary trunk originating from the truncus arteriosus gives rise to both pulmonary arteries. When both pulmonary arteries separate from the truncus arteriosus, with no vestige of a main pulmonary artery, they may arise close to one another (type II) or at some distance from one another

(type III). The type IV truncus arteriosus is now considered to represent a form of pulmonary atresia with ventricular septal defect (10). In 1965 Van Praagh and Van Praagh have proposed an expanded classification system that also includes two commonly associated abnormalities of the great arteries (11). Their type A1 corresponds to type I of Collett and Edwards, and type A2 encompasses type II and III. Type A3 includes cases with absence of truncal origin of one pulmonary artery, with blood supply to that lung from the ductus arteriosus or from a collateral artery. Type A4 is associated with underdevelopment of the aortic arch, including tubular hypoplasia, discrete coarctation, or complete interruption (12).

In our case the patient had truncus arteriosus communis type A2 by Van Praaghs which is characterized by separate but proximate origins of the left and right pulmonary arterial branches from the posterolateral aspect of the common arterial trunk. Presumably, this patient survived at birth because the pulmonary vascular resistance remained high and this prevented an excessive flow of blood into the pulmonary circulation directly from the persistent truncus (7). As a result of chronic exposure of the pulmonary vasculature to systemic arterial pressure, hypertensive pulmonary vascular disease may develop (12). In mild and moderate pulmonary hypertension there is thickening of

the muscular media and intimal fibrosis, as in this case. Severe pulmonary hypertension leads to irreversible changes such as angiomatoid and plexiform lesions (13). Recidiving hemoptysis and intraparenchymal pulmonary hemorrhage resulted from the anomalous anatomic origin of the pulmonary arteries. If there is massive cardiac hypertrophy, chronic subendocardial myocardial ischemia may develop, as in this case. Secondary polycythemia in this patient developed as a compensatory response to general tissue hypoxia caused by congenital cardiovascular anomaly. Extracardiac anomalies, present in 21 % to 30 % of autopsy cases of truncus arteriosus, include skeletal deformities, hydronephrosis, bowel malrotation, and multiple complex anomalies (12). We believe that hydronephrosis and hydronephrosis on the left side without any source of obstruction present in this case were extracardiac anomalies associated with truncus arteriosus. Surgical correction of truncus arteriosus in Slovakia wasn't available at the time this patient was diagnosed with the cardiovascular malformation. The cause of death in this case was cardiac failure.

CONFLICT OF INTEREST

The authors declare that there is no conflict of interest regarding the publication of this paper.

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