

Uhl's anomaly

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Abstract

Uhl's anomaly is characterized by absence of the myocardial layer of the right ventricle, with apposition of the endocardium and epicardium. It has often been confused with arrhythmogenic right ventricular dysplasia. Uhl's anomaly is rarely familial. It appears to be of congenital origin but is rarely associated with other cardiac malformations. Congestive cardiac failure is the usual mode of presentation, but arrhythmias or heart block are sometimes observed. Uhl's anomaly is exceedingly rare and it is not possible to estimate its prevalence. A review in 1993 found only 84 reported cases in the world literature since the beginning of the 20th century. Etiology has not been clearly determined yet. Diagnosis is generally suggested by echocardiography or more sophisticated methods of imaging. Medical management of any congestive heart failure or arrhythmias relies on palliative care but surgery, including cardiac transplantation, offers the only hope of extended survival.

Keywords

Uhl's anomaly, arrhythmogenic right ventricular dysplasia, parchment heart.

Disease name

Uhl's anomaly

Excluded disease

Arrhythmogenic right ventricular dysplasia (ARVD)

Definition

Uhl's anomaly is characterized by complete or partial absence of the myocardium of the right ventricle. The condition was named after Henry Uhl who reported the first case in 1952 (1). However, the "parchment heart" described earlier by William Osler in 1905 (2) may have been of the same nature. In 1979, Fontaine and

his colleagues (3) described a condition which they called "arrhythmogenic right ventricular dysplasia" (ARVD), characterized by local deficiency or fibro-fatty replacement of the right ventricular myocardium. A review of almost 500 cases in the literature up to 1993 (4) revealed that a variety of descriptive names were embraced by these two apparently distinct conditions, which included ectasia of the right ventricle, congenital aplasia of the right ventricle, congenital hypoplasia of the right ventricle, idiopathic right ventricular myocardial dysplasia, absence of the right ventricular myocardium, fatty infiltration and lipomatosis.

European Pediatric Cardiac Code

Reference of Uhl's anomaly is 07.01.06.

Clinical signs***Morphology and pathology***

There is virtually complete absence of the myocardium of the parietal wall of the right ventricle which is composed of apposing endocardial and epicardial surfaces with no interposed adipose tissue or evidence of inflammation or necrosis. In contrast the septal component, together with the septo-marginal trabeculation and the papillary muscles of the tricuspid valve, are normally muscularized. Localized discrete areas of similar morphology in the parietal wall have been recorded as examples of "partial Uhl's anomaly" as incidental findings at autopsy in a man of 66 years (5) and in a woman of 84 years (6).

In arrhythmogenic right ventricular dysplasia there is patchy and localized replacement of the muscle of the partial wall of the right ventricle by fibro fatty tissue which separates the endocardial and epicardial layers.

Main symptoms

Congestive cardiac failure is by far the most frequent symptom (4). It may be associated with massive peripheral edema (7) or massive pleural effusion leading to cardiac tamponade (8). It may rarely mimic other congenital cardiac malformations such as Ebstein's anomaly (9) or functional pulmonary atresia (10). Arrhythmias and conduction disturbances are not a predominant feature of Uhl's anomaly, probably due to absence of residual foci to initiate or transmit anomalous electrical activity. That it is why it differs from arrhythmogenic right ventricular dysplasia where palpitations, syncope, ventricular tachycardia, heart block or sudden death (often exercise-related) are the usual modes of presentation (4).

Associated anomalies

Pulmonary atresia with intact ventricular septum is sometimes associated with thinning and dilatation of the wall of the right ventricle, but this is probably a secondary manifestation. Apart from this, occurrence of other cardiac malformations in Uhl's anomaly is only fractionally higher than the overall incidence of congenital cardiac malformations in live births and can be explained on the basis of chance occurrence (4).

Incidence

Uhl's anomaly is exceedingly rare and it is not possible to estimate its prevalence. A review in 1993 found only 84 reported cases in the world literature since the beginning of the 20th century

(4). Patients' age at death ranged from one day to 84 years with a medium of 15 years and both sexes were equally affected. Arrhythmogenic right ventricular dysplasia was much more frequent, the medium age was higher (median 33 years) and men were affected more than twice as often as women.

The vast majority of reported cases have been sporadic but there have been some instances of familial occurrence, generally in siblings (11). Familial involvement in more than one generation is more likely to occur in arrhythmogenic right ventricular dysplasia (12).

Etiology and pathogenesis

Absence of right ventricular myocardium may be the result of primary non-development of myocytes in that region or may be due to selective apoptosis (16,17). The underlying cause may be genetic, either through parental transmission of a defective gene of autosomal recessive type (18), or of autosomal dominance with incomplete penetrance (19). Most cases are sporadic and may possibly be due to mutations or the results of exposure to toxic or infective agents. It has been suggested that overexpression of vascular endothelial growth factor, possibly by cardiomyocytes, may be responsible for the impaired development of the ventricular myocardium (20).

Diagnosis

Uhl's anomaly can hardly be missed at *post mortem* examination, but increasing use of echocardiography and other forms of scanning have facilitated accurate diagnosis during life and during fetal life (9). The typical clinical manifestations of congestive cardiac failure are not specific.

Treatment

Palliative treatment for congestive failure consists of standard forms of medication, including anti-arrhythmic drugs when indicated. Direct drainage of massive effusions into the pericardial, pleural or peritoneal cavities, and also from the legs, may be sometimes necessary (7,8). Successful surgical repair has been reported. Three types of procedures have been used. The most frequent approach has been exclusion of the right ventricle by closure of the tricuspid valve orifice with atrial septectomy and a bi-directional Glenn shunt (13). A modification of this procedure combines the atrial septectomy and bi-directional Glenn shunt with a partial right ventriculectomy, the so-called "one-and-a-half ventricular repair" (14). Successful complete correction by cardiac transplantation has also been reported (15).

References

1. Uhl, HS. A previously undescribed congenital malformation of the heart: lowest??? total absence of the myocardium of the right ventricle. *Bulletin of the Johns Hopkins Hosp* 1952;91:197-209.
2. Osler, WM. *The principles and practice of medicine*, 6th ed. New York. D. Appleton 1905;280.
3. Fontaine, G, Guirandon, G, Frank, R. Mechanism of ventricular tachycardia; surgical management based on epicardiac mapping. In: Narula OS, ed. *Cardiac arrhythmias*. Baltimore and London; Wilkins and Wilkins, 1979;516-523.
4. Gerlis, LM, Schmidt-Ott, SC, Ho, SY, Anderson, RH. Dysplastic conditions of the right ventricular myocardium: Uhl's anomaly & arrhythmogenic right ventricular dysplasia. *Br Heart J*. 1993;69:142-150.
5. Gould, L, Guttman, B, Carrasco, J, Lyon, AF. Partial absence of the right ventricular musculature. A congenital lesion. *Am J Med* 1967;42:636-641.
6. Sugiura, M, Hayashi, T, Ueno, K. Partial absence in the right ventricular muscle in an aged. *Jpn Heart J* 1970;11:582-585.
7. Kilinc, M, Akademir, M, Sivasli, E. A case of Uhl's anomaly presenting with severe right heart failure. *Acta Cardiol*. 2000;55:367-369.
8. Kisacik, HL, Ozdemir, K, Altunkeser, B, Oguzham, A, Goksel, S. Uhl's anomaly. *Jpn Heart J* 1999;40:503-507.
9. Benson, CB, Brown, DL, Roberts, DJ. Uhl's anomaly of the heart mimicking Ebstein's anomaly in utero. *J. Ultrasound Med*. 1995;14:781-783.
10. Tumbarello, R, Adatia, I, Yetman, A, Boutin, C, Izukawa, T, Freedom, RM. From functional pulmonary atresia to right ventricular restriction. Long term follow up of Uhl's anomaly. *Int J Cardiol* 1998;67:161-164.
11. Diggleman, U, Baur, HR. Familial Uhl's anomaly in an adult. *Amer J Cardiol*, 1984;53:1402-1403.
12. Laurent, M, Descaves, C, Biron, Y, Deplace, C, Almange, C, Daubert, J. Familial form of arrhythmogenic right ventricular dysplasia. *Am Heart J* 1987;113:827-829.
13. Azhari, N, Assaqqat, M, Bulbul, Z. Successful surgical repair of Uhl's anomaly. *Cardiol Young* 2002;12:192-195.
14. Yoshii, S, Suzuki, S, Hosaka, S, Osawa, H, Takahasi, W, Takizawa, K, Abraham, SJ, Tada, Y, Sugiyama, H, Tan, T, Kadono, T. A case of Uhl's anomaly treated with a one and a half ventricle repair combined with partial right ventriculectomy in infancy. *J. Thorac Cardiovasc Surg* 2001;122:1026-1028.
15. Ikari, NM, Azeka, E, Aiello, VD, Atik, E, Barbero-Marcial, M, Ebaid, M. Uhl's anomaly. Differential diagnosis and indications for cardiac transplantation in an infant.
16. James, TN, Nichols, MM, Sapir, DW, D. Patre PL, Lopez, S.M. Complete heart block and fatal ventricular failure in an infant. *Circulation* 1996;93:1588-1600.
17. Uhl, HS. Uhl's anomaly revisited. *Circulation* 1996;93:1483-1484.
18. Nava, A, Thiene, G, Canciani, B, Scognamiglio, R, Daliento, L, Buja, G, Martini, B, Stritoni, P, Fasoli G. Familial occurrence of right ventricular dysplasia: a study involving nine families. *Am Coll Cardiol* 1988;12:1222-1228.
19. Ruder, MA, Winston, SA, Davis, JC, Abbot, JA, Eldar, M, Scheinmann, MM. Arrhythmogenic right ventricular dysplasia in a family. *Am J Cardiol* 1985;56:799-800.
20. Feucht, M, Christ, B, Wilting, J. VEGF induces cardiovascular malformation and embryonic lethality. *Amer J Path.* 1997;151:1407-1416.