PICTORIAL REVIEW / Cardiovascular imaging

MR imaging of arrhythmogenic right ventricular dysplasia: What the radiologist needs to know


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KEYWORDS
Arrhythmogenic right ventricular dysplasia; Cardiac MRI; Cardiomyopathy

Abstract  Arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C) is an inherited heart muscle disease that predominantly affects the right ventricle. Clinical manifestations are related to severe ventricular arrhythmia that may lead to sudden death, mostly in young patients. Magnetic resonance imaging (MRI), included in the new diagnostic criteria since 2010, aims to detect segmental and global wall motion abnormalities, reduced ejection fraction, right ventricular dilatation and right ventricular diastolic/systolic dysfunction. An MRI assessment of the right ventricle is often challenging, partly because the MRI diagnostic criteria have some limitations, and also because it requires a significant learning curve due to the low prevalence of the disease. Therefore, this article aims to review the pathophysiology of the disease, the cardiac MRI protocol, images of the various stages of this affection as well as the differential diagnosis.

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Arrhythmogenic right ventricular dysplasia (ARVD) or arrhythmogenic right ventricular cardiomyopathy is an inherited cardiomyopathy characterized by fibro-fatty replacement, mainly affecting the right ventricle (RV) but also the left ventricle (LV). In 50% of the cases, this genetic disease is inherited in an autosomal dominant pattern with mutations in the genes coding for desmosomal proteins. ARVD causes severe ventricular

Abbreviations: ARVD, Arrhythmogenic right ventricular dysplasia; MRI, Magnetic Resonance Imaging; RV, Right ventricle; LV, Left ventricle; ECG, Electrocardiogram.
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Arrhythmias in young individuals and is responsible for sudden death. There is no pathognomonic feature of ARVD, thus diagnosis is based on a combination of clinical, electrical and morphological criteria, as reported in the literature [1]

Franck and Fontaine first described the disease in 1978. Initial diagnostic criteria followed in 1994. Recent genetic findings as well as advances in cardiac imaging, especially in MRI, have led experts to update the criteria in 2010, by adding cardiac MRI to the imaging modalities [1].

MRI assessment of the disease is challenging for the following reasons:
- the right ventricle is functionally totally different from the left ventricle;
- a high number of artifacts on cine sequences (SSFP) due to the anterior position of the RV against the chest wall, thereby compromising the quality of the findings that may be misinterpreted for dyskinesia;
- RV volume and ejection fraction are difficult to measure and very dependent on the loading conditions;
- frequent artifacts related to the detection of associated signs such as fat infiltration and retention of contrast in the myocardial wall.

This article reviews:
- what radiologists must know about ARVD;
- how to perform an MRI assessment;
- how to measure the ejection fraction of the RV using commercially available equipment;
- what signs to look for to diagnose ARVD;
- what are the other disorders causing similar signs, and differential diagnosis.

Clinical and pathophysiological characteristics

ARVD is a genetic cardiomyopathy. The inheritance pattern is usually autosomal dominant with variable penetrance and expression, and, but far less often, autosomal recessive. The incidence of the familial form of ARVD is estimated between 15 and 50% [2]. The majority of the mutations are in genes coding for desmosomal proteins (desmoplakin, plakoglobin, plakophilin-2, desmoglein-2, and desmocollin-2) involved in the intercellular connection between myocytes. Desmosomal dysfunction is believed to cause myocytes to detach from each other, leading to cell death, inflammation and repair by fibro-fatty substitution, and finally RV dysfunction and dilatation [3].

The prevalence of ARVD is estimated between 1:2000 and 1:5000. There is a male predilection, with a male-to-female ratio 3-1 [2]. ARVD is a major cause of sudden death in athletes. The first symptoms generally occur between the second and fifth decades of life (median age: 29 years). The presenting symptoms include palpitations, syncope and sudden death in decreasing order of frequency. The incidence of sudden death decreases after the fourth decade [2], but incidence of heart failure in ARVD patients has been reported to follow in 20% of the cases. Annual mortality rate is 2.3% and the mean age at death is 54 ± 19 years. The primary causes of death are heart failure and sudden death. A diagnosis of ARVD makes family screening necessary to detect subclinical manifestations of the disease; and genetic testing may be considered [3].

Diagnostic criteria

ARVD is difficult to diagnose. It is based on clinical and paraclinical criteria, including family history. The diagnostic criteria were revised in 2010 and are subdivided into major and minor criteria (Table 1). A ''definite'' diagnosis consists of 2 major criteria or 1 major and 2 minor criteria, or else 4 minor criteria. A ''borderline'' diagnosis consists of 1 major and 1 minor criteria or 3 minor criteria. Finally a ''possible'' diagnosis consists of 1 major or 2 minor criteria. The criteria were updated by including RV functional abnormalities detectable by echocardiograms, MRI and angiography, histological evidence from endomyocardial biopsy specimens, electrocardiographic abnormalities (repolarization, conductivity, and arrhythmia), and information from family history [1].

Thus, MRI must be part of a comprehensive evaluation of the disease and be based on specific clinical, electrocardiographic and arrhythmic criteria, as defined by the Marcus diagnostic criteria.

Other morphological abnormalities such as fat infiltration and delayed enhancement images, are detectable by MRI, but have not been included in the diagnostic criteria. These abnormalities are described here and their detection is useful in case of wall motion abnormalities.

MRI

MRI is performed while the patient is in supine position, with an imaging system that comprises a dedicated cardiac coil and ECG monitoring. ECG-synchronized cardiac MRI images are acquired during breath-holding in the following planes: 4-chamber, short axis, RV and LV 2-chamber views and infundibular view (Fig. 1). Fig. 1 shows the axes for the analysis of the RV; the LV axes are widely described in the literature.

In some patients, MRI assessment may be hindered or prevented by artifacts caused by extrastoles. There are two ways to overcome this. The first way is to administer a beta-blocking antiarrhythmic drug (5mg Atenolol by slow intravenous injection). For a patient with extrastoles despite a long-term beta-blocking treatment, we use Atropine (1 mg diluted into 10 mL saline serum injected over a period of 2 min) to increase cardiac rate, and restore a regular heart rate. Such a treatment may not be initiated in case of polymorphic extrastoles. Another possibility is to use SSFP sequences with arrhythmia rejection.

Protocol

Cine imaging

Steady-State-Free-Precession multi shot sequences (FIESTA, true-FISP, Balanced Fast Field Echo): T2/T1 weighted sequences, where fluids appear bright (e.g. bright blood hypersignal). They are less sensitive to flow-related...
<table>
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<th>Table 1</th>
<th>Arrhythmogenic right ventricular dysplasia (ARVD) diagnostic criteria according to the 2010 Revised Task Force Criteria.</th>
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<tr>
<td><strong>Major criteria</strong></td>
<td><strong>Minor criteria</strong></td>
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<tr>
<td><strong>Structural dysfunctions and abnormalities</strong></td>
<td><strong>Echocardiography</strong></td>
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<tr>
<td><strong>MRI</strong></td>
<td>Regional RV akinesia or dyskinesia or dyssynchronous RV contraction And one of the following RV end-diastolic volume index ≥ 110 mL/m² (male) or ≥ 100 mL/m² (female) Or RV ejection fraction ≤ 40%</td>
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<tr>
<td><strong>Angiography</strong></td>
<td>Segmental akinesia or dyskinesia or aneurysm</td>
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<td><strong>Tissue characterization, histology</strong></td>
<td>Residual myocytes &lt; 60% by morphometric analysis (or &lt; 50% if estimated), with fibrous replacement of the RV free wall myocardium in ≥ 1 sample, with or without fatty replacement on endomyocardial biopsy</td>
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<td><strong>Depolarization and conduction abnormalities</strong></td>
<td>Inverted T waves in right precordial leads (V1, V2 and V3) or beyond in individuals &gt; 14 years of age (in the absence of complete right bundle-branch block with QRS ≥ 120 ms)</td>
</tr>
<tr>
<td><strong>Arrhythmias</strong></td>
<td>Sustained or nonsustained ventricular tachycardia with left bundle-branch morphology with superior axis (negative or indeterminate QRS in leads DII, DIII and VF and positive in lead VL)</td>
</tr>
<tr>
<td><strong>Family history</strong></td>
<td>ARVD confirmed in a first-degree relative who meets Task Force criteria ARVD confirmed pathologically at autopsy or surgery in a first-degree relative Identification of pathogenic mutation categorized as associated or probably associated with ARVD in the patient under evaluation</td>
</tr>
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artifacts. These sequences provide good contrast between blood pool and myocardium showing clear delineation of endocardial borders. Therefore, they are used for the assessment of the segmental and global ventricular dysfunction (ejection fraction and volume measurement) as well as for the anatomic assessment of the cardiac chambers [4]. These imaging sequences are essential to detect RV dysplasia, since they are able to evidence the diagnostic criteria. Therefore, we use them as first-line imaging modality.

The views are four-chamber, short axis, LV and RV long axis and RV infundibulum. In our group, we use the 4-chamber view to visualize the entire right ventricle (5 slices). Slice thickness is 6 mm, with repetition time (TR) ≤ 35 ms, minimum time to echo (TE), and 36 to 40 cm field of view (FOV) [5].

Morphologic sequences

Black-blood single shot fast spin echo sequences reduce motion artifacts and shorten the imaging time, but at the cost of loss of visualization of the RV wall [4].

These sequences are useful to identify intramyocardial fat infiltration.

Delayed contrast enhancement sequence (inversion recovery gradient echo)

This sequence is acquired 10 to 15 min after IV injection of gadolinium chelates (0.2 mL/kg). Images can be acquired using 2D or 3D techniques, but require specific adjustment of the inversion time of the right ventricle.

Sequences are performed in the 4-chamber and short-axis views.

Phase-sensitive Inversion-Recovery sequences generate images that are not sensitive to inversion time artifacts [4].

Optimal protocol selection

SSFP cine sequences are the most important ones to evidence signs of ARVD. In our group, we perform the complete set of cine imaging. Only in case of abnormal cine sequences, do we acquire T1 and delayed enhancement images.

RV ejection fraction

MRI is currently considered the Gold Standard for measuring ventricular function; its excellent spatial resolution has proven to be a specific and reproducible tool to evaluate LV and RV anatomy, volume and myocardial mass. This is particularly true for the RV since data provided by echocardiographic and isotopic techniques may be difficult to interpret especially when the RV has an abnormal shape or size.

Simpson’s rule method is used (just like for the LV) [6]. Infundibulum’s rule must be included in the measurement because it can account for 25 to 30% of RV volume. Two rules to accurately contour and measure the RV are:

- to determine the most basal level of the RV and to separate it from the right atrium in the short-axis images. Short- and long-axis cine sequences are helpful for this;
- to perform an endocardial contouring with inclusion of the RV trabeculae in the ventricular cavity at end-diastole and systole (Fig. 2).

MRI findings

Wall motion and functional abnormalities

Unlike the initial diagnostic criteria, the 2010 updated criteria are quantitative.

Only three types of MRI-detectable abnormalities have been included in the criteria to diagnose ARVD: segmental RV wall motion abnormalities, RV dilatation and reduction of ejection fraction.

Intramyocardial fat infiltration and delayed enhancement have not been included.

A major criterion detectable by MRI is the combination of regional RV akinesia or dyskinesia or dyssynchronous RV
contraction with one of the following: ratio of RV end-diastolic volume to body surface area $\geq 110 \text{mL/m}^2$ in male patients and $\geq 100 \text{mL/m}^2$ in female patients or RV ejection fraction $\leq 40\%$.

A minor criterion is the combination of regional RV akinesia or dyskinesia or dysynchronous RV contraction with one of the following: $100 \leq$ RV volume at end-diastole $< 110 \text{mL/m}^2$ in male patients and $90 \leq$ Volume at end-diastole $< 100 \text{mL/m}^2$ in female patients or $40\% <$ RV ejection fraction $\leq 45\%$ [1].

Reduction of the ejection fraction
Segmental wall motion abnormalities and RV dilatation modify the RV ejection fraction.

When associated with segmental wall motion abnormalities, an RV ejection fraction $\leq 40\%$ is considered a major criterion, and a minor criterion if between 41 and 45%.

Right ventricle dilatation
The evolution of the RV volume is key in monitoring the progression of the disease [7]. RV dilatation may be segmental, typically involving the infundibulum, or the basal or else mid-ventricular segment of the free wall, or may be global (Fig. 3). The diagnostic sensitivity and specificity of ventricular dilatation for ARVD is 77% and 95–100%, respectively [7].

Wall motion abnormalities
The detection of focal contraction abnormalities is more subjective than the measurement of RV volume and ejection fraction. MRI interpretation of the right ventricle and the RV free wall in the strict axial view can produce many artifacts, due to magnetic field inhomogeneities, causing false-positive readings of dyskinesia. The artifacts are less visible on the 4-chamber sequences but local shimming is sometimes required to avoid them (Fig. 4). We highly recommend the use of 4-chamber views for the assessment of RV wall motion abnormalities [8].

The wall motion abnormalities to evaluate are [7]:
- hypokinesia where an area or segment of the ventricular wall is characterized by reduced systolic wall thickening or shortening (wall thickening $< 40\%$) (Fig. 5; Cine 1);
- akinesia: systolic wall thickening $< 10\%$ (Fig. 6; Cine 2);
- dyskinesia: abnormal outward movement during systole (Fig. 7; Cine 3);
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Figure 3. 57-year-old man with diagnosed arrhythmogenic right ventricular dysplasia (ARVD). Right ventricle (RV) global hypokinesia with marked dilatation of the RV and secondary right atrial dilatation. The left ventricle (LV) volume is normal without any wall motion abnormality: a: SSFP multi shot sequence, 4-chamber view, at end-diastole; b: SSFP multi shot sequence, 4-chamber view, at end-systole; c: SSFP multi shot sequence, short-axis view at end-diastole; d: SSFP multi shot sequence, short-axis view at end-systole; e: SSFP multi shot sequence, RV long-axis view, at end-diastole; f: SSFP multi shot sequence, RV long-axis view, at end-systole.

• diastolic bulging or bulging: dyskinetic area during systole with aneurysm during diastole (Fig. 8; Cine 4 and 5).

These functional abnormalities often correlate with areas of signal abnormality visible on black blood sequences. Abnormal wall motion associated with black blood signal abnormality are more suggestive of ARVD than either abnormality alone [7]. These segmental wall motion abnormalities usually affect the basal and mid ventricular segments of the RV free wall with an incidence of 70—80%. Specificity reaches 100% if these dyskinetic areas show segmental dilatation of the RV.

On the other hand, false-positive signals of bulging at end of diastole in individuals free of ARVD are possible. These artifacts are seen at the apical third of the RV free wall.

Figure 4. a: 4-chamber view showing an artifact at the apex of the right ventricle (RV) (broad arrow), in front of the RV wall (small white arrows); b: we note the absence of the artifact on the sequence in the same 4-chamber view, but with a slightly different radiofrequency (open arrow).
Figure 5. 46-year-old man resuscitated from “sudden death” without any history of cardiomyopathy. Functional analysis shows global hypokinesia of the right ventricle (RV) free wall; a: SSFP multi shot sequence 4-chamber view at end-diastole; b: SSFP multi shot sequence 4-chamber view at end-systole.

Morphological abnormalities

Intramyocardial fat infiltration

Intramyocardial fat has long been considered a major MRI diagnostic criterion, however today it is considered less reliable.

Epicardial fat infiltrates from the epicardium to the endocardium of the myocardial wall, causing a disruption in the clear line of demarcation between the myocardium and the epicardial fat (Figs. 9 and 10). Fat usually appears as an intramyocardial hyperintense spin-echo T1 signal. Fat infiltrates into the following areas: infundibulum, RV free wall, trabeculae, RV moderator band and right side of the ventricular septum.

The prevalence of intramyocardial hyperintense signals in ARVD patients ranges between 22 to 75% in different studies.

Figure 6. 10-year-old child with suspected arrhythmogenic right ventricular dysplasia (ARVD). Imaging shows several segmental wall motion abnormalities: akinesia of basal free wall segment (full arrow), area of dyskinesia on the mid-free wall (arrowhead) and hypokinesia of the RV apex (dotted arrow); a: SSFP multi shot sequence, short-axis view, at end-diastole; b: SSFP multi shot sequence, short-axis view, at end-systole; c: SSFP multi shot sequence, 4-chamber view at end-diastole; d: SSFP multi shot sequence, 4-chamber view at end-systole.
Moreover, there is poor inter-observer reproducibility for the analysis and MRI detection of fat due to:
• the presence of epicardial and pericardial fat, which makes identification of intramyocardial fat challenging;
• the subtricuspid area may be difficult to distinguish from the atrioventricular groove, which is rich in fat;
• the RV free wall is only 3–5 mm; the spatial resolution of cardiac MRI is therefore often not so good;
• the normal presence of fat deposits in healthy individuals.

These deposits are observed in healthy individuals in 15% of the cases, most often in the anteroapical region of the right ventricle and in up to 50% in overweight individuals. The prevalence is also higher in older people. Some experts recommend correlating these intramyocardial hyperintense signals with segmental wall motion.
abnormalities to separate normal fat from pathological fat infiltration.

Because of these difficulties, intramyocardial fat identification by MRI was not included in the diagnostic criteria of 2010 [1]. Fat infiltration is an additional argument that may confirm diagnosis when associated with dilated ventricle or with contractile dysfunction. Since fat infiltration is not considered a diagnostic criterion for ARVD, it must not be systematically looked for in the absence of RV functional, volume or contractility abnormality.

Wall thickness

Several authors have reported cases of ARVD associated with a thinning, or even a thickening, of the RV wall [7]. However, the prevalence varies a lot depending on the study. These abnormalities are never isolated but always appear in combination with segmental wall motion abnormalities. These signs have not been included in the international recommendations [1].

Hypertrabeculation

The incidence of hypertrabeculation has been reported 40% in several studies, with hypertrophied structures, such as papillary muscles and moderator band (Fig. 11). This finding has been equated to an initial angiographic finding that had a “stack of plates” appearance. However, this abnormality is not specific for ARVD and may be present in any condition that results in RV hypertrophy or dilatation, and should therefore not be retained as a criterion.

Delayed enhancement

Myocardial delayed enhancement images correspond to areas of fibrosis, inflammation or edema, where the extracellular volume is increased. In delayed enhancement images, retention of gadolinium remains visible in the pathological areas, while it is washed out of normal myocardium (Figs. 12–14; Cine 6 and 7).

The use of delayed enhancement in ARVD has been assessed and was shown to have excellent correlation with histological findings and inducible ventricular arrhythmias by electrical stimulation. Furthermore, there is a strong association between the extent of delayed enhancement and RV dysfunction. The prevalence is estimated to be 67% [9].
Figure 12. Cardiac magnetic resonance imaging (MRI) in asymptomatic 54-year-old man with family history of arrhythmogenic right ventricular dysplasia (ARVD). The different sequences show delayed enhancement in the basal free wall (arrow head) and the apex (full arrow) with hypokinetic areas, as well as delayed contrast enhancement in the mid free wall (dotted arrow) with a dyskinetic area with fat infiltration; a: SSFP multi shot sequence, 4-chamber view at and-diastole; b: SSFP multi shot sequence, 4-chamber view at end-systole; c: TSE T1 “Black-blood” sequence 4-chamber view; d: delayed enhancement 4-chamber view; e: delayed enhancement 4-chamber view, slightly different slice level; f: delayed enhancement short axis.

Figure 13. 30-year-old man with suspected arrhythmogenic right ventricular dysplasia (ARVD) based on wall motion and conductivity abnormalities. Subepicardial delayed contrast enhancement in the right ventricle (RV) free wall with hypokinetic (full arrow) and normokinetic (dotted arrow) areas; a: SSFP multi shot sequence, short-axis view, at end-diastole; b: SSFP multi shot sequence, short axis, at end-systole; c: delayed enhancement short axis.

However, delayed enhancement has not been incorporated in the diagnostic criteria when they were revised in 2010, because of the difficulties to assess the RV and its thin wall [1].

Delayed enhancement remains however helpful to confirm diagnosis when the fibrotic area is located in an area involved by contractile abnormality. It will thus be useful to look for, if the sequences used for the functional analysis suggest ARVD.

Left ventricular involvement

Left ventricular involvement in ARVD has been observed for many years, ever since the first descriptions of the disease. Prevalence has been reported between 16 and 76% of the patients, depending on the study. Fibro-fatty replacement can affect either diffusely or regionally; affect the septum and/or, more often, the free wall, with a predilection for postero-septal and postero-lateral areas. Involvement is subepicardial, midwall, or transmural and extends from the epicardium towards the endocardium. MRI-detected morphological abnormalities are identical to those observed in the RV and delayed enhancement may be observed unrelated to the wall motion abnormalities [10] (Fig. 15).

The prevalence of LV dysfunction is higher in patients with definite ARVD diagnosis than in patients with doubtful ARVD. The segmental wall motion abnormalities, like for the RV, appear before the modification of the ejection fraction. MRI “tagging” may increase the sensitivity of the diagnosis.

LV involvement has, traditionally been considered a late stage manifestation of the progressive disease, however, this has recently been brought into question with cases
ventricular tachycardia (or "Gallavardin infundibular ventricular tachycardia"). The initial examination consists of Holter monitoring, electrocardiogram, echocardiography and sometimes a stress test and/or in some cases examination for late ventricular potentials. If results are inconclusive, an MRI may be performed. Given the frequency of isolated infundibular systoles in the general population and the low incidence of ARVD, an MRI is not warranted in each patient as a first-line investigation.

Catecholaminergic polymorphic ventricular tachycardia

Genetic channelopathy with familial transmission is characterized by severe polymorphic ventricular arrhythmias induced by adrenergic stress (physical effort, sport). The arrhythmias occur in children and young adults without underlying heart disease and cause convulsion, syncope and/or sudden death. The differential diagnosis of ARVD may be based on the clinical signs. The MRI findings are normal and the typical stress test results confirm the diagnosis.

Sarcoidosis

Cardiac involvement is often part of a multi organ involvement of this disease but is also, in some rare cases, limited to the single cardiac site. The most common sites of sarcoid granulomatous infiltration are located in the basal interven- tricular septum, and the LV anteroseptal wall.

Several cases of isolated RV involvement with clinical and imaging findings highly suggestive of ARVD have been described. RV involvement in sarcoidosis can occur as a dilatation and dysfunction of the RV, with focal akinesia, dyskinesia or aneurysm raising the question of the differential diagnosis of ARVD.

Myocarditis

Cases of myocarditis mimicking ARVD have been reported. They are characterized by selective involvement of the right ventricle causing functional and structural abnormalities such as microaneurysms.

Differential diagnosis

Right ventricular outflow tract tachycardia

Ventricular extrasystoles originating in the infundibulum, due to abnormal automaticity with triggered activity and without underlying heart disease, are considered benign. The condition may consist of bursts of infundibular

Figure 14. (Same patient than in Fig. 13). Delayed enhancement of hypokinetic areas at level of infundibulum (dotted arrow) and right ventricle (RV) free wall (full arrow) and normokinetic area corresponding to the RV apex (arrowhead); a: SSFP multi shot sequence, RV long-axis view, at end-diastole; b: SSFP multi shot sequence, RV long-axis view, at end-systole; c: delayed enhancement 4-chamber view; d: delayed enhancement RV long-axis view. evidencing LV involvement without RV involvement, supporting the adoption of a new term, "arrhythmogenic cardiomyopathy" [10].

Figure 15. 40-year-old woman with suspected arrhythmogenic right ventricular dysplasia (ARVD) based on wall motion abnormalities and family history of ARVD. Left ventricle (LV) involvement is evidenced by subepicardium and midwall delayed contrast enhancement in the inferior and lateral walls (arrow); a: delayed enhancement short-axis view; b: delayed enhancement short axis, different slice.
Moreover, histological evidence of myocarditis has often been evidenced in endomyocardial biopsies from patients with ARVD, pointing to the hypothesis that the disease may have an inflammatory etiology. Also, some viruses (adenovirus, enterovirus, coxsackievirus) are known for their higher cardiac tropism in ARVD patients. The role of these viruses in the pathophysiology of ARVD is not yet known, and particularly whether they contribute to the development of the disease or whether a dysplasia-affected myocardium is more prone to virus infection. Nevertheless, viruses should be considered a significant environmental factor with probably a key role in the development of the disease.

Uhl anomaly

Uhl anomaly, a very rare congenital heart disease (84 cases were described in the literature up to 1993), was first described in 1952. It is characterized by the absence of right ventricular free wall myocardium, with normal interventricular septum and LV wall.

Endocardium and pericardium are directly apposed, with no interposed adipose tissue, which makes the RV look like “parchment”. The tricuspid valve has a normal shape and hinges normally.

Clinical signs appear in childhood with both genders equally affected, and without any significant family history. The disease progresses rapidly, and is often fatal in the absence of cardiac transplant. Symptoms include dyspnea, cyanosis, and edema in lower limbs due to right ventricular failure. The etiology is unknown but may be related to apoptosis.

The MRI images depict an extremely thin-walled RV, due to the absence of myocardium, associated with progressive RV dilatation. The right atrium is dilated and hypertrophied due to congestive heart failure.

Conclusion

Cardiac MRI is frequently performed in patients with suspected ARVD. However, the number of diagnosed cases is low. Despite the update of the diagnostic criteria in 2010, the specificity of the ARVD diagnosis remains high, but its sensitivity has remained unchanged. Diagnosis is still challenging whatever the imaging modality used. MRI is the most highly reproducible and specific technique for assessing the morphology and segmental wall motion of the right ventricle. The assessment remains highly observer-dependent and requires a deep knowledge of MRI images of normal RV as well as those of abnormalities suggestive of ARVD. Multidisciplinary management is mandatory for:

- correctly prescribing cardiac MRI;
- handling first-degree relatives screening;
- initiating treatment and appropriate clinical follow-up.

Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at [doi:10.1016/j.j.diiii.2014.07.009].

Disclosure of interest

The authors declare that they have no conflicts of interest concerning this article.

References