Uhl Anomaly

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Uhl anomaly is a very rare condition characterized by complete or partial absence of the right ventricular myocardium, which is replaced by fibroelastic tissue [1].

The true incidence of Uhl anomaly is unknown, but less than 100 confirmed cases have been described in the literature [2]. The cause is thought to be a high apoptotic activity, which begins during the perinatal period or early in infancy, leading to destruction of the right ventricular wall [2, 3]. Histologic examination reveals partial or total absence of the myocardium of the parietal wall of the right ventricle and direct apposition of the opposing endocardial and epicardial surfaces [4, 5]. This leads to thinning of the right ventricular free wall. Uhl anomaly usually presents in neonates or infants as right-sided heart failure. Patients rarely survive to adulthood. No effective treatment other than heart transplant has been shown to improve survival.

Computed tomography (CT) findings include an extremely thin-walled right ventricle with complete or partial absence of the right ventricular free-wall myocardium and a paucity of trabeculations (Figs. 8.1 and 8.2) [6, 7]. The tricuspid valve, interventricular septum, and left ventricular myocardium are normal.

Uhl anomaly needs to be distinguished from arrhythmogenic right ventricular dysplasia. Histologically, the latter disorder is characterized by patchy replacement of right ventricular myocardium with fibrofatty tissue, primarily occurring within the ventricular outflow tract and inlet or apical regions. Arrhythmogenic right ventricular dysplasia usually produces ventricular arrhythmias and manifests during adolescence, whereas Uhl anomaly presents in infancy with heart failure. CT shows fatty infiltration of the right ventricular free wall in arrhythmogenic right ventricular dysplasia which may help to differentiate it from Uhl anomaly, which has a paucity of myocardium and apical trabeculations as well as an absence of fatty infiltration.

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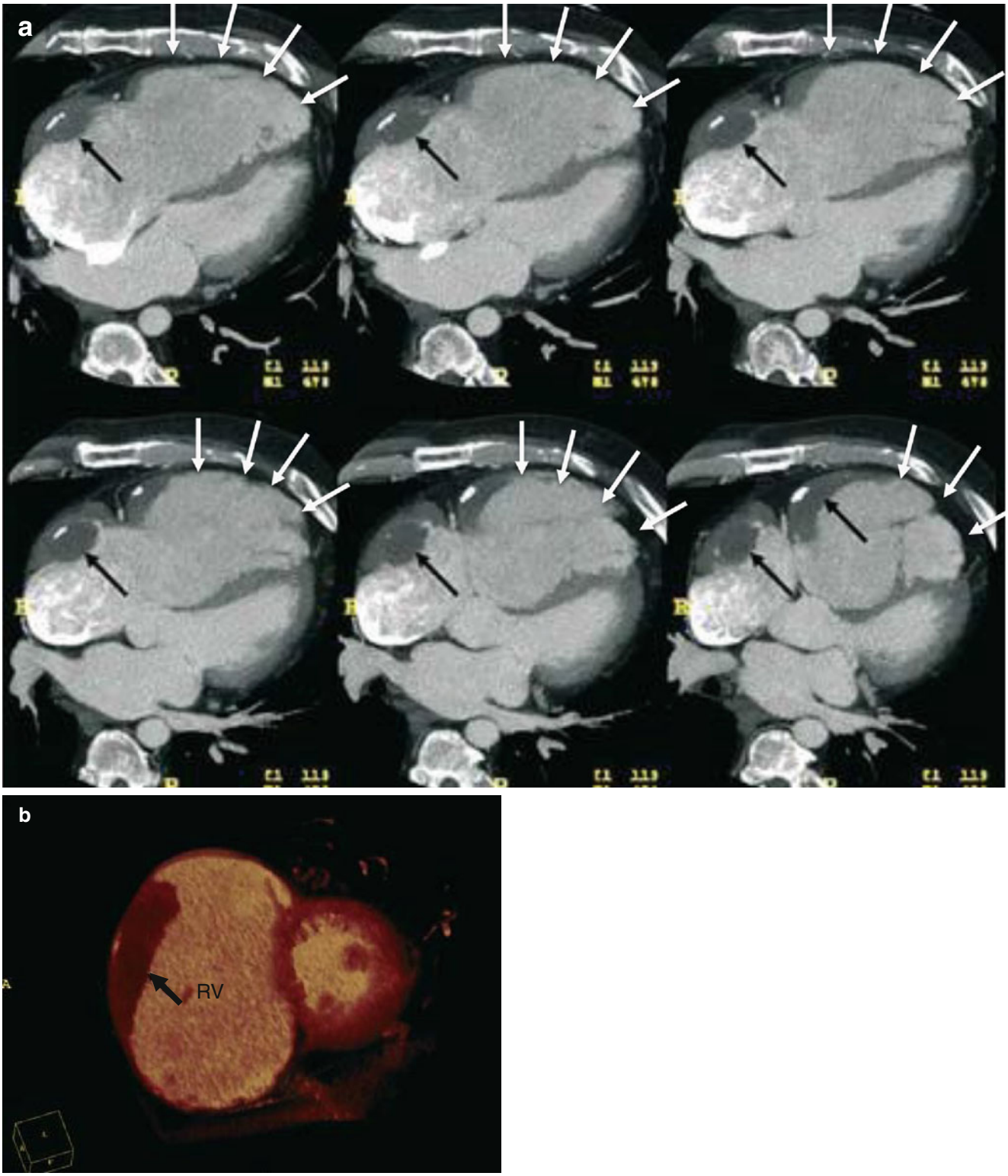

Fig. 8.1 Partial Uhl anomaly in a 51-year-old man. Panel (**a**) is an axial image and panel (**b**) is a short-axis image. Both panels show partial absence of the right ventricular wall (*white arrows*). Note the massively dilated right ventricle (*RV*) and right atrium. Chronic mural thrombus with foci of calcifications is seen (*black arrows*). (Reproduced from Cheng et al. [7]. With kind permission of Springer-Verlag, Berlin Heidelberg, Germany)

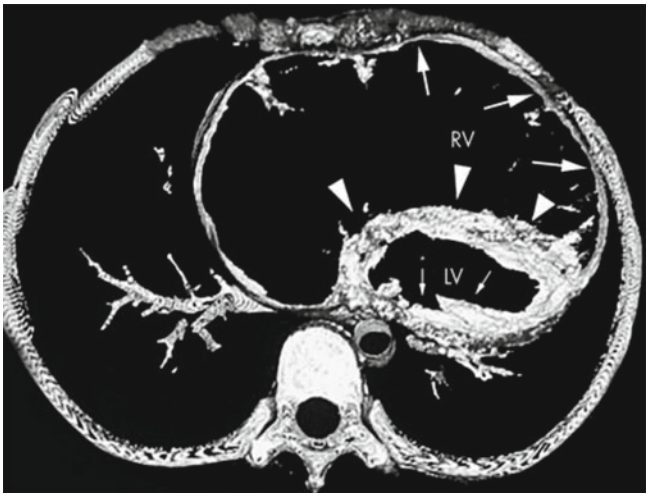

Fig. 8.2 Uhl anomaly. An axial scan obtained by a multisliced computed tomography scanner which dramatically illustrates an extremely dilated right ventricle (RV) with almost a complete absence of RV myocardium (*thin arrows*). The interventricular septal myocardium (*arrowheads*) is normal thickness (Reproduced from Ceviz et al. [6]. With kind permission from BMJ Publishing Group Ltd)

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