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IMAGE FOCUS

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Multimodality imaging of neonatal rhabdomyoma and follow-up under sirolimus treatment

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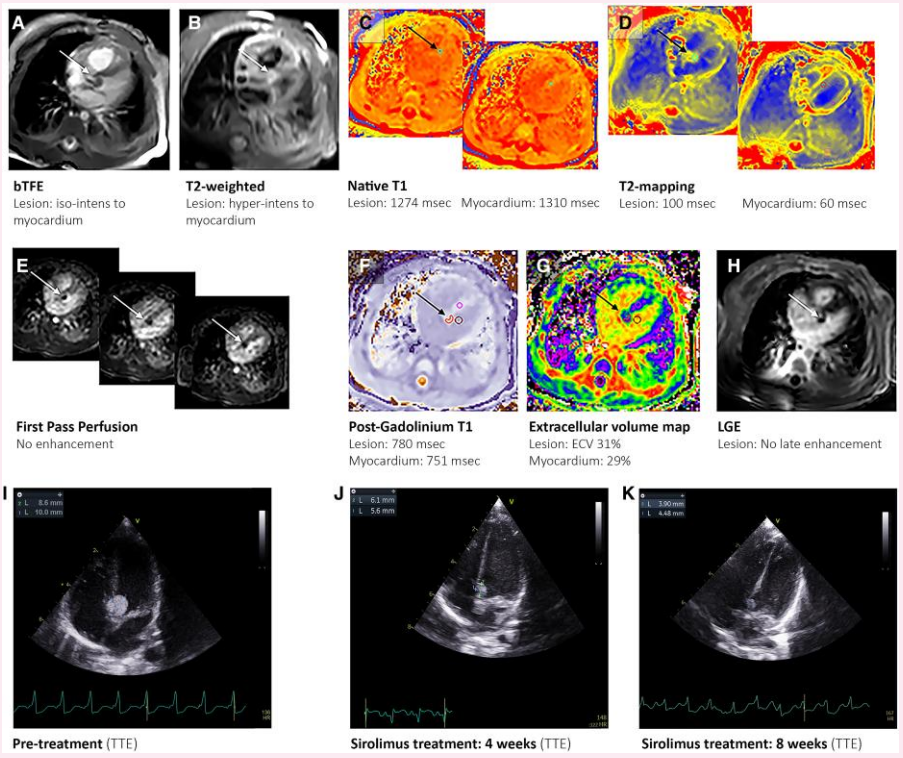
A 3-day-old neonate (birth weight 4440 g) was referred for evaluation of a systolic murmur detected at birth. Transthoracic echocardiography revealed a 10.0 × 8.6 mm solitary mass in the left ventricular outflow tract (LVOT), originating from the interventricular septum below the aortic valve. The mass partially obstructed the LVOT, with a Doppler velocity of 2.0 m/s.

Differential diagnoses included rhabdomyoma, fibroma, and other cardiac tumours. Cardiac tumours in children are rare (0.01–0.32%), with rhabdomyoma being the most common neonatal cardiac tumour (~60%). Rhabdomyoma raises concern due to its strong association with tuberous sclerosis complex (TSC), a genetic disorder caused by mutations in the TSC1 or TSC2 genes, which regulate cell growth and proliferation. These mutations lead to benign tumours, or hamartomas, in multiple organs, including the brain, skin, kidneys, heart, and lungs. Although ~80% of children with rhabdomyomas also have TSC, the solitary presentation of the tumour and the absence of typical TSC features in this neonate warranted further evaluation with cardiac magnetic resonance (CMR) imaging.



CMR under general anaesthesia, utilizing a foetal cardiac imaging protocol (3 Tesla, ECG-gated, no breath-hold, 3 averages, semi-3D), revealed normal cardiac anatomy with a 7 × 5 × 9 mm solitary lesion causing LVOT narrowing. The lesion showed iso-intensity on bTFE, normal native T1 values, hyperintensity on T2-weighted imaging, slightly elevated T2 mapping values (~100 ms), no first-pass perfusion, no late gadolinium enhancement, normal post-gadolinium T1 values, and normal extracellular volume (*Panels A–H*). Findings supported the diagnosis of rhabdomyoma. Cranial MRI was normal.

Sirolimus treatment significantly reduced tumour mass and LVOT gradient over 8 weeks (*Panels I–K*, Supplementary data online, *Videos S1–S3*), supporting the diagnosis of rhabdomyoma. Post-treatment recurrence occurred, but LVOT gradients remained low, requiring no additional therapy. Genetic testing for TSC was negative.

While histology remains the gold standard, CMR is highly effective in suggesting aetiology and guiding management. CMR provided valuable insights with specific MR characteristics, complementing echocardiography in assessing tumour size, location, and haemodynamics, even in neonates. Thereby, invasive diagnostics can be avoided.

Supplementary data are available at *European Heart Journal - Cardiovascular Imaging* online.

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