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#### **INTRODUCTION & OBJECTIVES**

Duchenne muscular dystrophy (DMD) is a X-linked devastating recessive disorder characterized by progressive muscle weakness, affecting boys. This debilitating primarily condition is caused by mutations in the dystrophin gene, leading to the absence or deficiency of the dystrophin protein essential for muscle function. As the disease progresses, individuals experience mobility challenges, respiratory complications, and cardiac issues, significantly impacting their quality of life.

We present two patients with ischemic stroke, a relatively rare complication in DMD and different risk factors.

Showing typical symptoms, both patients received an MRI examination that revealed an occlusion of the right middle cerebral artery, a thrombectomy and anticoagulation afterwards. Fortunately both recovered very well without any neurological deficits due to the ischemic stroke.

The diagnostic workup of the first patient (13 years) revealed a severely reduced left ventricular function with an ejection fraction (EF) below 10%, due to a dilatative cardiomyopathy (DCMP) on echocardiography and MRI. After frustrating heart failure therapy, the patient received a left ventricular assist device (LVAD). CASE 2 The other patient (24 years) showed a milder DCMP with an EF of 30%, but additionally the heterozygous factor V Leiden mutation and the 4G/5G polymorphism of the plasminogen activator inhibitor type 1 gene were detected.



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# Ischemic stroke in two patients with Duchenne muscular dystrophy and different risk factors

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# CASE 1

Figure: 13-year-old (a) with a proximal occlusion of the M1 segment of the right middle cerebral artery (red arrow) and 24-year-old (**b**) with a diffusion-perfusion mismatch volume of 22ml (ratio = 2,1)

# REFERENCES



### CONCLUSION

These two cases highlight different risk factors for strokes in patients with DMD. The DCMP, a major complication should be monitored and treated according to the standards of care. A predisposition to an increased risk of thrombosis may also play a role in the pathogenesis of strokes in DMD patients, but is not routinely investigated.

Stroke in DMD patients is rare, but has an enormous impact on the patient's quality of life if additional residual symptoms persist. For this reason, larger studies on this topic would be desirable in order to obtain a valid stroke risk assessment, including coagulation testing. And, as a next step, the possibility of prophylactic anticoagulation in patients with multiple risk factors should be evaluated, to prevent further deterioration of mobility and quality of life.

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