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The genetic connection

Case report

A young Caucasian male with long standing severe debilitating muscle weakness and chronic respiratory failure (ventilator dependant) was transferred to the Intensive care unit for management of ventilator associated pneumonia. A cardiology evaluation was sought in view of clinical and radiographic signs of heart failure. Family history was positive for a similar problem in one of his siblings. The patient's ECG revealed the following characteristic findings (fig. 1). What is the probable diagnosis?

Discussion

In our patient the ECG findings were classic for dystrophinopathy (Duchenne's and Becker's muscle dystrophy). Tall R wave (20 mm) in V₁ and prominent Q waves in lateral leads (I, aVL, V₃–V₆) were noted. Other notable findings included right axis deviation, right bundle branch block (RBBB) pattern and R in V₁ significantly greater than r'. R in V₁ is considered to be dominant if R/S >1 (R >7 mm typically). In the presence of RBBB pattern R >r' and R >10 mm are considered pathologic. Echocardiogram revealed global hypokinesia, LVEF of 30%, no regional wall motion abnor-

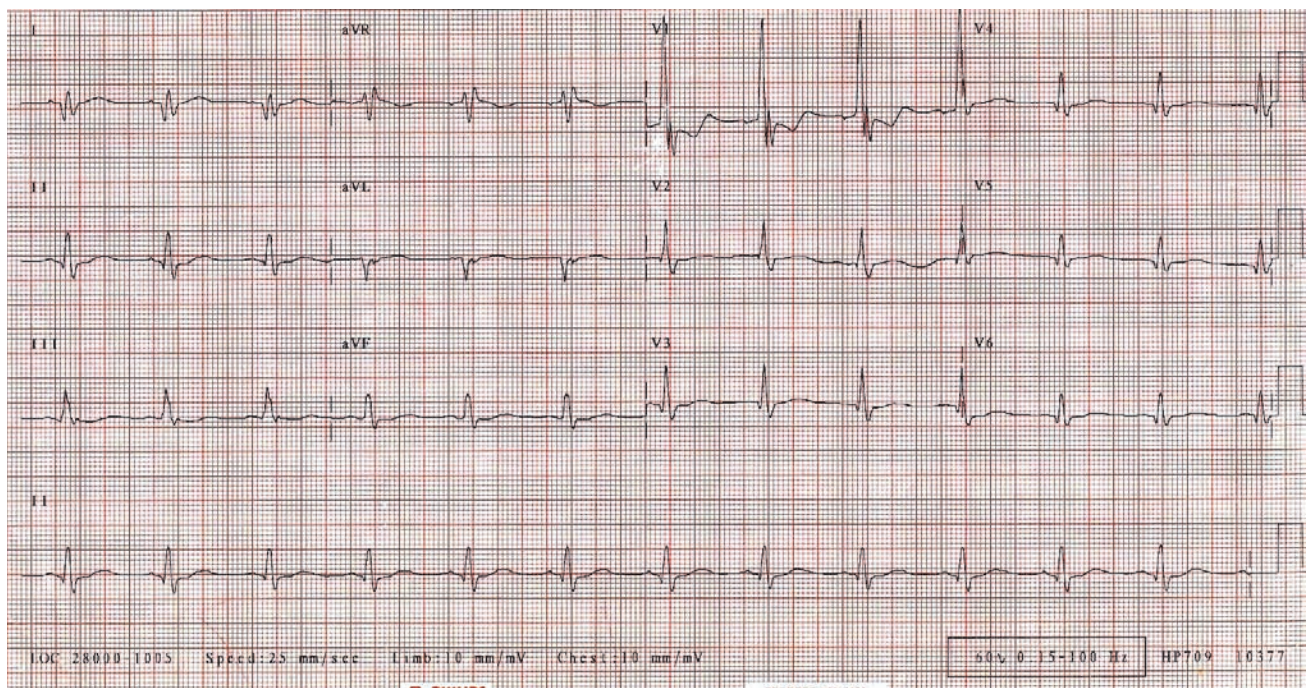


Figure 1
Admission ECG showing the classic tall R wave in V₁.

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There is no conflict
of interest.

Table 1
Differential diagnosis of tall R in V₁.

Posterior myocardial infarction
Hypertrophic cardiomyopathy
Right ventricular hypertrophy
Acute RV strain (pulmonary embolism)
Ventricular pre excitation
Dextroposition/dextrocardia Normal variant

malities, valvular abnormalities and normal right ventricular function. Review of patient's prior records confirmed the diagnosis of Duchenne's muscle dystrophy. Table 1 lists the various causes for tall R in V₁. This patient was treated with conventional medical therapy for heart failure.

Cardiac involvement is extremely common in patients of dystrophinopathy. 70–90% have an abnormal ECG. Sinus tachycardia is the most common abnormality. Tall R waves in V₁ and deep Q waves in the left lateral and inferior leads are considered to be characteristic of dystrophinopathies [1]. Following the early descriptions by Manning and Cropp numerous series have corroborated the occurrence of this distinctive pattern of QRS changes [2]. In one series prominent R in V₁ was noted in 88%, prominent Q wave in lateral leads in 73% and 37% in inferior leads. This abnormality is thought to represent selective early scarring of

the posterobasal and lateral wall of left ventricle as a result of relatively greater stress consequent to the longitudinal myocyte orientation [1, 2]. Other abnormalities include intraventricular conduction delays, non-specific ST-T wave changes and changes in PR and QT intervals. Increased ectopy, bradyarrhythmias and heart blocks are known but infrequent. In the presence of RBBB these patients usually demonstrate Rsr' or RSR' patterns in contrast to the usual rsR' pattern. It is important to note that ECG changes have been documented even in the absence of clinical or echocardiographic cardiac involvement and even in asymptomatic female carriers. Members of the same family often have similar ECG abnormalities. The ECG changes are independent of the severity and progression of skeletal muscle disease and therefore do not carry prognostic significance (in contrast to LVEF) [3].

References

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