PICU admission following unexplained cardiac arrest

A 16-year-old male was admitted to the PICU following unexplained cardiac arrest after experiencing ventricular fibrillation while running in gym class. He had a longstanding history of neurodevelopmental and medical concerns, including hypotonia, autism spectrum disorder, global developmental delay, and mild pulmonary artery stenosis, with years of prior subspecialty care and nondiagnostic genetic evaluations.

3 years old

Initial outpatient genetic evaluation occurred at age 3.

Prior genetic testing included chromosomal microarray, Fragile X testing, and an autism/ intellectual disability multi-gene panel – all of which were nondiagnostic.

Ongoing care

Over the course of childhood, the patient had **more than 700 visits** to the hospital for subspecialty care and therapies.

16 years old

While running in gym class, the patient experienced ventricular fibrillation cardiac arrest.

Inpatient genetics was consulted in the PICU due to his unexplained cardiac arrest and prior medical history. Notably, this hospital had recently implemented a policy change allowing broader, first-line rapid genome sequencing to be ordered in the PICU.

5 days later

Results from the rapid genome sequencing identified a *DMPK* triplet repeat expansion consistent with myotonic dystrophy.

Within hours of the diagnosis, the following changes to medical management were made:

1. Cancellation of same-day procainamide challenge for evaluation for Brugada syndrome
2. Change of type of device implantation to internal cardiac defibrillator
3. Avoidance of propofol by anesthesia team
4. Urgent same-day pulmonary consult revealed previously unrecognized respiratory insufficiency due to neuromuscular weakness and BiPAP during sleep was initiated
5. Post-operative recovery was planned for the PICU rather than the cardiac floor
6. Screening TSH, hemoglobin A1C, and CK were obtained
7. His mother was referred for adult cardiac evaluation due to her own risk, which subsequently detected cardiomyopathy requiring ICD implantation
8. Accurate recurrence risk counseling was performed

By removing barriers to ordering rapid genome sequencing as a first-tier test, the patient finally received a precise diagnosis that explained his condition and *immediately impacted* clinical management.

References: Keefe AC. Scott AA. Kruidenier L. et al. Am J Medical Genetics. 2026. 0:1-11

Photo does not represent actual patient.