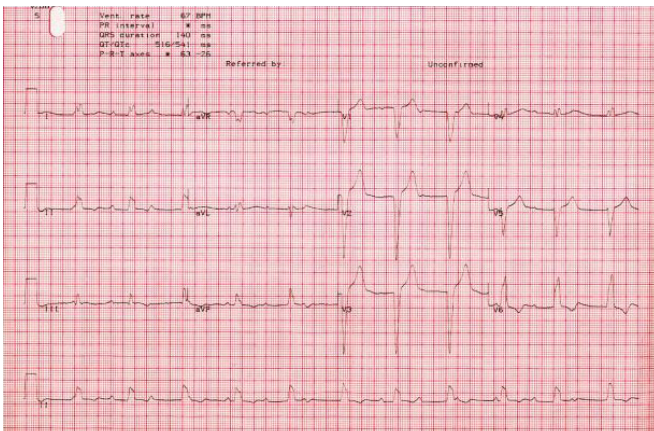


Case 1

Following a syncopal episode at home, this 39-year-old man suffered from a fractured clavicle. He admitted to being breathless on effort and complained of difficulty in walking and poor vision. His facial appearance and his electrocardiogram (ECG) are shown below.



Q

Questions:

1. What three features are shown in the picture?
2. What might explain his poor vision?
3. What is the clinical diagnosis? How is this condition inherited?
4. Which chromosome is abnormally affected?
5. What is the ECG diagnosis?
6. What precise treatment should he receive?

A

Answers:

1. Frontal balding, bilateral ptosis and long, haggard expressionless features.
2. Cataracts and external ophthalmoplegia.
3. Dystrophia myotonica. Autosomal dominant.
4. Chromosome 19.
5. Complete heart block.
6. Dual chamber permanent pacemaker implantation.