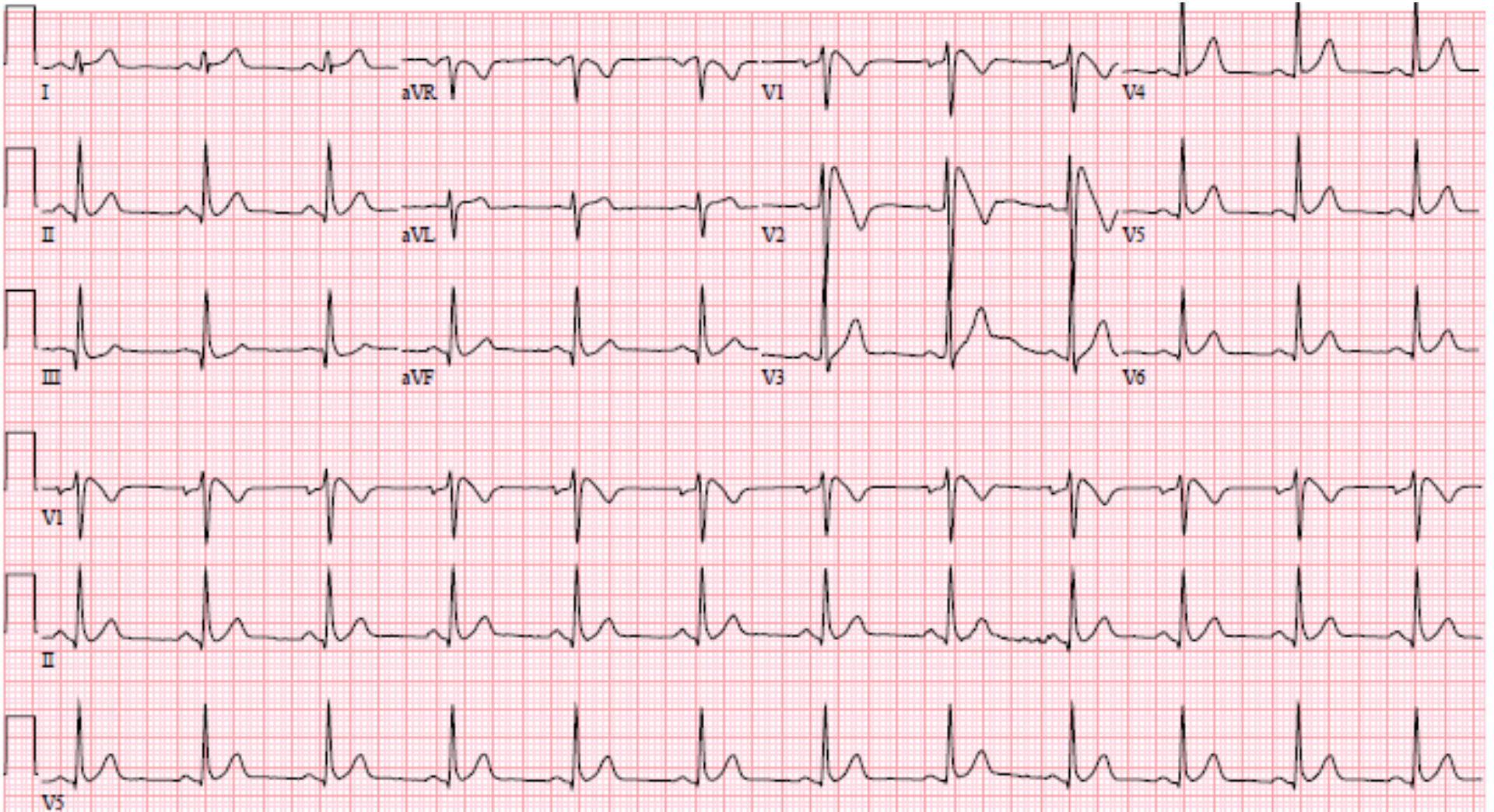
A 9 year old girl comes into your primary care clinic for routine evaluation. Her mother mentions that the girls' father was recently diagnosed with some sort of heart problem, but she cannot recall what it is. She does, however, have a copy of her husband's ECG, seen below.

1) What is the major abnormality on this ECG? Don't name the diagnosis, just describe the abnormality. (1 point)

2) What is the diagnosis? Hint—this problem is a "channelopathy" that raises the risk of sudden cardiac death (1 point)

3) What are the chances that your 9 year old patient has the same diagnosis (from a genetic standpoint, that is)? (1 point)



1) What is the major abnormality on this ECG? Don't name the diagnosis, just describe the abnormality. (1 point)

ST segment elevation in leads V1 and particularly lead V2. More specifically, the ST elevation has a distinct down-sloping character. This is NOT "J-pointing" or early repolarization, where the ST segment has a "J" shaped intersection with the QRS complex.

2) What is the diagnosis? Hint—this problem is a "channelopathy" that raises the risk of sudden cardiac death (1 point)

This ECG is very consistent with an entity known as Brugada syndrome. It is caused by a mutation in one of the important sodium channels in the cardiac myocytes responsible for myocardial depolarization (most channelopathies affect channels that control repolarization). Interestingly, the same channel is implicated in long QT syndrome type 3. Individuals with Brugada syndrome are at elevated risk of sudden cardiac death from the spontaneous development of VT/ Torsades de Pointe, with degeneration into V-fib.

3) What are the chances that your 9 year old patient has the same diagnosis (from a genetic standpoint, that is)? (1 point)

Brugada syndrome as well as many other channelopathies are inherited in an autosomal dominant fashion. Therefore, your patient has a 50% chance of having the same mutation. However, penetrance is estimated at about 30%, and ECG abnormalities are seen in about 80% of individuals who have the Brugada mutation, so chances are your patient will have a normal ECG when screened.

For more information on this rare but very interesting disease, visit:
http://www.ncbi.nlm.nih.gov/books/NBK1517/?log$=disease8_name