

## Learning Points

1. We identified a novel mutation in the *HPGD* gene, c.468T>A, leading to a change in the amino acid histidine to glutamine (p.His156Gln).
2. Mutations in *HPGD* can be associated with the triad of keratoderma, hyperhidrosis and clubbing.

## Question 1

1. The *HPGD* gene encodes for an enzyme that is involved in which of the following?
  - a) Interleukin anabolism
  - b) Interleukin catabolism
  - c) Prostaglandin anabolism
  - d) Prostaglandin catabolism
  - e) Prostaglandin expression

## Question 2

2. Based on the involvement of the *HPGD* gene in the pathogenesis of palmoplantar keratoderma, hyperhidrosis and digital clubbing, which of the following may be used in the treatment of this condition in the future?
  - a) Leukotriene analogues
  - b) Leukotriene inhibitors
  - c) Prostacyclin analogues
  - d) Prostaglandin analogues
  - e) Prostaglandin inhibitors

## Answers to question 1

- a) Incorrect, the enzyme is not involved in interleukin anabolism
- b) Incorrect, the enzyme is not involved in interleukin catabolism
- c) Incorrect, the enzyme is not involved in prostaglandin anabolism
- d) Correct, the *HPGD* gene encodes for an enzyme involved in prostaglandin catabolism
- e) Incorrect, the enzyme is not involved in modulating prostaglandin expression

## Answers to question 2

- a) Incorrect, leukotriene analogues are not a potential treatment for this condition
- b) Incorrect, leukotriene inhibitors are not a potential treatment for this condition
- c) Incorrect, prostacyclin analogues are not a potential treatment for this condition

- d) Incorrect, prostaglandin analogues are not a potential treatment for this condition
- e) Correct, prostaglandin inhibitors theoretically may pose as a possible therapeutic option for this condition