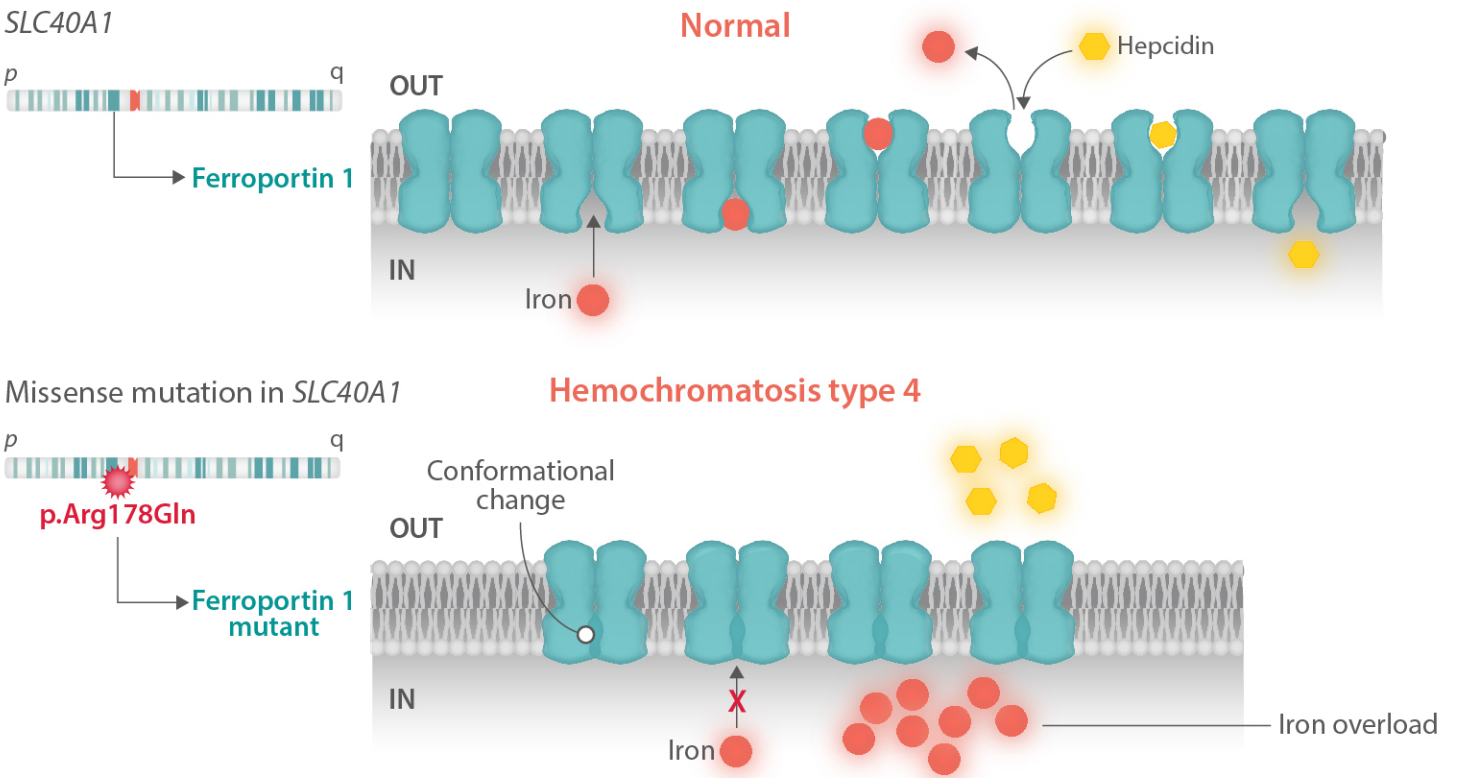
The p.Arg178Gln missense mutation in *SLC40A1* is associated with a novel pathogenic mechanism of hemochromatosis type 4



- p.Arg178Gln missense mutation represents a new category of loss-of-function mutations and is recurrent in the SLC40A1 gene of patients showing typical reticuloendothelial iron overload
- p.Arg178Gln ferroportin 1 mutant shows reduced ability to export iron out of the cell

Ka *et al.*, Haematologica, 2018