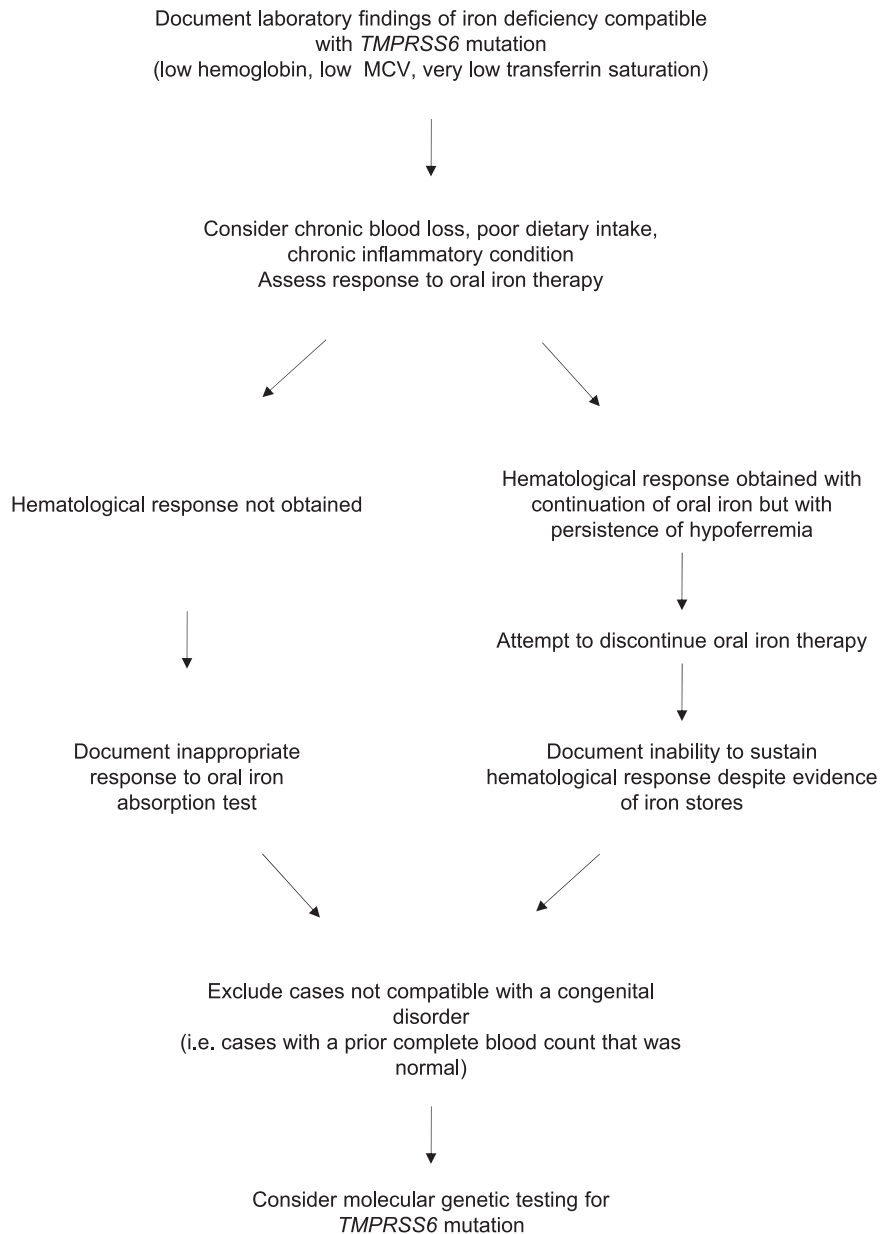


Supplemental Information

SUPPLEMENTAL TABLE 4 List of Genes Involved in Iron Metabolism Further Analyzed Using WES Data Obtained on Both Patients

Gene	Full gene name	Protein
<i>ABCB7</i>	ATP-binding cassette, sub-family B (MDR/TAP), member 7	ABCB7
<i>ALAS2</i>	Delta-aminolevulinate synthase 2	ALAS2
<i>BMP6</i>	Bone morphogenetic protein 6	BMP6
<i>CYBRD1</i>	Cytochrome b reductase 1	CYBRD1
<i>FTH1</i>	Ferritin, heavy polypeptide 1	Ferritin H chain
<i>FTL</i>	Ferritin, light polypeptide	Ferritin L chain
<i>FXN</i>	Frataxin	Frataxin
<i>GDF15</i>	Growth differentiation factor 15	GDF15
<i>GLRX5</i>	Glutaredoxin 5	Glutaredoxin 5
<i>GPI</i>	Glucose-6-phosphate isomerase	Glucose-6-phosphate isomerase
<i>HAMP</i>	Hepcidin antimicrobial peptide	Hepcidin
<i>HFE</i>	Hemochromatosis	HFE
<i>HFE2</i>	Hemochromatosis type 2	Hemojuvelin
<i>HMOX1</i>	Heme oxygenase (decycling)1	Heme oxygenase-1
<i>HMOX2</i>	Heme oxygenase (decycling)2	Heme oxygenase-2
<i>MON1A</i>	MON1 homolog A	Vacuolar fusion protein MON1 homolog A
<i>NEO1</i>	Neogenin	Neogenin
<i>SLC11A2</i>	Solute carrier family 11, member 2	DMT1
<i>SLC25A28</i>	Solute carrier family 25, member 28	SLC25A28
<i>SLC25A37</i>	Solute carrier family 25, member 37	Mitoferrin-1
<i>SLC39A1</i>	Solute carrier family 39, member 1	ZIP1
<i>SLC40A1</i>	Solute carrier family 40, member 1	Ferroportin
<i>SMAD4</i>	SMAD family member 4	SMAD4
<i>STEAP3</i>	STEAP family member 3	STEAP3
<i>TF</i>	Transferrin	Transferrin
<i>TFR2</i>	Transferrin receptor 2	TfR2
<i>TFR1</i>	Transferrin receptor 1	TfR1
<i>TWGS1</i>	Twisted gastrulation homolog 1	TWGS1



SUPPLEMENTAL FIGURE 1

Suggested diagnostic algorithm for the evaluation of IRIDA.