G6PD Genotyping Algorithm for Therapeutic Drug Recommendations

**G6PD** / Glucose-6-Phosphate Dehydrogenase (G6PD) Full Gene Sequencing

- **Male (hemizygous)**
  - WHO Class I Variant
    - Positive
    - Deficient with CNSHA phenotype
  - WHO Class II or III Variant
    - Positive
    - Deficient
  - WHO Class IV Variant
    - Normal

- **Female (heterozygous)**
  - WHO Class I Variants
    - Positive
    - Deficient with CNSHA phenotype
  - WHO Class I and II or III Variants
    - Positive
    - Deficient, at risk for CNSHA phenotype
  - WHO Class II or III Variants
    - Positive
    - Deficient
  - WHO Class IV Variants
    - Normal

- **Female (homozygous/compound heterozygous)**
  - WHO Class IV and I, II, or III Variants
    - Indeterminate phenotype

**Therapeutic Recommendation:**
- **Contraindication** for drugs associated with hemolytic anemia in G6PD-deficient patients
  - See: Pharmacogenomic Associations Tables

- **G6PD Normal**
  - Therapeutic Recommendation: No G6PD-related contraindication of drugs/compounds

CNSHA: Congenital nonspherocytic hemolytic anemia

*Variants of Uncertain (VUS) significance may be identified. If a VUS is identified, a WHO Class will not be assigned and enzyme studies are recommended.

1 WHO Class I Variant: Severe G6PD enzyme deficiency (<10% activity) and chronic nonspherocytic hemolytic anemia (CNSHA)
2 WHO Class II/III Variant: Deficient G6PD enzyme activity (10%-60% activity)
3 WHO Class IV Variant: Normal G6PD enzyme activity

<60% G6PD activity symptomatic G6PD deficiency carrier
>60% G6PD activity unaffected G6PD deficiency carrier

Indeterminate-G6PD enzyme assay needed
Order: G6PD / Glucose-6-Phosphate Dehydrogenase (G-6-PD), Quantitative, Erythrocytes

G6PD Deficiency Diagnosis

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