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| NORMAL ADULT HEMOGLOBIN | | | |
|---|--|---|---|
| HbA | Alpha ₂ Beta ₂ (97%) | | |
| HbA ₂ | Alpha ₂ Delta ₂ (2.5%) | Increased in Beta-thal trait | |
| HbF | Alpha ₂ Gamma ₂ (<1%) | Increased levels ameliorate sickle cell disease | |
| <u>Diseases</u> | <u>Genotype</u> | <u>Clinical</u> | <u>Diagnosis</u> |
| Alpha-Thal-2 | 1 Alpha-chain defective | Silent | DNA analysis, family history |
| Alpha-Thal-1 | 2 Alpha-chains defective | Microcytosis, mild anemia (mid 30's) | DNA analysis, family history |
| Hb H disease | 3 Alpha-chains defective | Microcytosis, hemolysis, splenomegaly, moderate anemia | DNA analysis, family history, Heinz body prep |
| Beta-thal minor (Beta ⁺ thal) | 1 Beta-chain defective | Microcytosis, target cells, mild-moderate anemia (30's) | Hb electrophoresis |
| Beta-thal major (Beta ^o) | 2 Beta-chains defective | Severe anemia, hemolysis | Hb electrophoresis |
| Beta-thal intermediate (Beta ⁻) | 2 Beta-chains defective with residual normal Beta-chain production | Variable between minor and major thal | Hb electrophoresis |
| Hb E trait | Alpha ₄ Beta ₁ E ₁ | Microcytosis, not anemic | Hb electrophoresis |
| Hb E disease | Alpha ₄ E ₂ | Microcytosis, target cells, not anemic | Hb electrophoresis |
| Sickle cell disease | HbS | Sickle cell disease | Hb electrophoresis |
| Sickle cell trait | HbSA | None | Hb electrophoresis |
| SC disease | HbS/HbC | Mild-mod sickle disease, target cells, splenomegaly | Hb electrophoresis |
| Sickle-thal | HbS/Beta-thal gene | Severe sickle disease with microcytosis | Hb electrophoresis |
| Alpha-thal/Beta-thal major | Alpha-thal/Beta-thal major | Beta-thal intermediate | Hb electrophoresis |
| HbE/Beta-thal | HbE/Beta-thal | Beta-thal major | Hb electrophoresis |