

## *GENETIC DISEASES AND PHARMACOGENOMICS*

### *Table of Contents*

- 1. Overview*
- 2. Nucleic Acid Amplification*
- 3. Chromosome Imaging*
- 4. Genomics Technologies*
- 5. Proteomics Technologies*
- 6. Current Pharmacogenomic Tests*
- 7. Future Pharmacogenomic Testing*
- 8. Major Diseases*
  - \* Achondroplasia*
  - \* Autosomal Dominant Polycystic Kidney Disease*
  - \* Cancer*
  - \* Cosmetogenomics*
  - \* Cystic Fibrosis*
  - \* Down's Syndrome*
  - \* Duchenne and Becker Muscular Dystrophy*
  - \* Factor V (Leiden)*
  - \* Factor IX Deficiency*
  - \* Fragile X Syndrome*
  - \* Heart Disease*
  - \* Hemochromatosis*
  - \* Hemophilia*
  - \* Huntington's Disease*
  - \* Maternal-Fetal Incompatibility*
  - \* Multiple Endocrine Neoplasia*
  - \* Phenylketonuria (PKU)*
  - \* Polycystic Kidney Disease (PKD)*
  - \* Prenatal Screening*
  - \* Retinitis Pigmentosa*
  - \* Retinoblastoma*
  - \* Sickle Cell Anemia*
  - \* Spinal Muscular Atrophy*
  - \* Vitamin B12 Metabolism*
- 9. Social Issues and Concerns*

*Table: Major Companies Developing or Marketing  
Molecular Diagnostic Tests For Genetic Diseases*