

Clinical Sequencing

Clinical NGS Applications

- Diagnosis of rare genetic diseases
- Cancer precision medicine and treatment selection
- Pharmacogenomics - drug response prediction
- Prenatal and newborn screening
- Infectious disease identification

Clinical Considerations

Quality Standards

- CLIA/CAP certification
- High coverage (>30X)
- Validated pipelines
- Quality control metrics

Interpretation

- ACMG variant classification
- Clinical significance
- Actionable findings
- Secondary findings reporting

Ethical Issues

- Informed consent
- Incidental findings

Reimbursement

- Insurance coverage
- CPT codes

- Data privacy
- Genetic counseling

- Medical necessity
- Prior authorization

Requires multidisciplinary team: clinicians, geneticists, bioinformaticians, counselors