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project baby deer

Considerations When Comparing Clinical Laboratories for Whole Genome Sequencing (WGS)

1. **Comprehensiveness of the test.**
   - ☐ What sample types are accepted for WGS?
     i. Blood, saliva, cord blood and/or dried blood spots?
   - ☐ What variant types are clinically validated by the laboratory?
     i. Mitochondrial DNA, copy number variants (CNVs), spinal muscular atrophy types 1 and 2 (SMA 1/2), single-nucleotide variants (SNVs) and/or insertion/deletion mutations?
   - ☐ What type of genetic disorders might not be detected by genome?

2. **Testing approach and price.**
   - ☐ Does the lab offer rapid turnaround time for proband only?
     i. Proband=patient sample.
     ii. Trio=patient sample + both parent samples.
   - ☐ Does the lab offer ultra-rapid turnaround time for critically sick patients?
   - ☐ What are the corresponding CPT codes and fee schedule prices for the laboratory's test options?

3. **Reporting**
   - ☐ What is the turnaround time for a final result report?
   - ☐ Does the laboratory provide a preliminary result report?
     i. If yes, what is the turnaround time?
   - ☐ How does the laboratory communicate results?
     i. Verbal/phone call?
     ii. Written communication through email or the lab portal?
   - ☐ Can the laboratory provide a sample positive report and a sample negative report?
   - ☐ How does the laboratory treat secondary/incidental findings?
     i. If incidental findings are identified in a parent but not in the proband, are those results reported?
   - ☐ Under what circumstances do you report variants of unknown significance (VUS) or candidate genes?
     i. If a VUS is changed in the future to likely pathogenic, does the lab notify families or the ordering physician?

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4. **Service offerings.**
    - □ Is your institution able to compliantly do business with the WGS clinical laboratory?
        i. Is a formal lab service agreement required to send testing?
        ii. Is your send-out lab willing to ship to the WGS lab or must the clinicians arrange to use and send pre-paid FedEx kits?
            1. This can impact billing workflows after the test is done.
    - □ Can the laboratory recommend any options for genetic tele-mentoring/tele-consult support services for clinical teams and families?
    - □ Does the laboratory offer access to the lab directors and/or genetic counselors?
        i. Are genetic counselors and/or laboratory directors available to discuss patient qualification for testing, review genomic results and provide clinical management guidance?
    - □ Does the laboratory offer family variant testing to at-risk members at no additional charge?
    - □ What is required from a system and process standpoint to order testing?
        i. Lab portal registration and access?
    - □ Where is genetic data stored?
    - □ How long is a patient's genetic data stored?
    - □ How can parents or clinical teams access the genetic data in the future?
    - □ What is the lab's policy about timing and cost of repeat analysis when testing is non-diagnostic?

For reference and access to glossary of terms, see: GeneReviews Glossary - GeneReviews® - NCBI Bookshelf (nih.gov)