Recommended Testing Algorithm

Test 1: Chromosome Breakage
- Negative Breakage Study: Patient does not have FA, or possible mosaicism
- Positive Breakage Study: Confirms diagnosis of FA

Test 2: Fanconi Anemia Panel (16 genes) by Next Generation Sequencing
- No mutations identified or 1 mutation identified in AR gene, or variant(s) of uncertain significance
- Two mutations identified in same AR gene or 1 mutation in FANCB: Genetic diagnosis of FA
- Deletion/Duplication testing

*Complementation Testing (available for research/investigational purposes only)

This is the suggested testing algorithm. Please note that any test can be requested in any order.
*Contact 513-636-5998 for details regarding complementation testing on a research/investigational basis.