Genetic diagnostic strategy in intestinal malabsorption of cobalamin

1. Exclude nutritional deficiency and parasites
2. Test for proteinuria; presence supports IGS but absence is not informative for IFD
3. Collect DNA samples from patient, parents, and siblings (if any)
4. Determine ethnicity
5. Priority 1 search for founder mutations in that ethnicity
6. Priority 2 search for other known mutations in that ethnicity
7. Priority 3 screen remaining exons in GIF, then AMN, and finally CUBN