

Fanconi Anemia Complementation Analysis Available

The CAP/CLIA Accredited Translational Trials Development and Support Laboratory of the Division of Experimental Hematology at the Cincinnati Children's Hospital Medical Center has received an NIH award to supplement the cost of testing for Fanconi Anemia Complementation Groups. The complementation assay is now available to clinicians and researchers for the cost of \$150 per sample (complementation analysis) plus \$180 (preparation of cell lines, if needed) for testing from either peripheral blood (EBV transformed lymphoblastoid cell lines – LCLs) or skin biopsies (fibroblast cell lines), as required. Contact Dr. Punam Malik (punam.malik@cchmc.org, 513-636-8588) or Lilith Reeves (lilith.reeves@cchmc.org, 513-636-3468) for more information or to schedule testing.

The Fanconi anemia (FA) Complementation assay allows reliable determination of FA complementation in patients with confirmed FA diagnosis by DEB or MMC for clinical and laboratory purposes using retroviral gene transfer correction of melphalan-induced cell cycle arrest (see Chandra et al. Molecular Therapy, 2005). The service is partially funded through an NIH Grant (R01HL081499-01A, DA Williams) and is available to clinicians and researchers at a subsidized rate of \$150 per sample plus the cost for establishing cell lines from peripheral blood or skin biopsies, if necessary. The assay is CAP/CLIA-compliant and SOP-driven, allowing use of the data in a clinical setting and for clinical trial enrollment decisions. It also facilitates targeted sequencing for mutation analysis. Instructions are also provided for participation in the International Fanconi Anemia Registry (IFAR), which facilitates subsequent mutation identification, also in a CLIA-compliant laboratory. For patients entering the IFAR, no charges are incurred for cell line derivation, complementation or mutation determination.

The FA Complementation assay is an efficient method for the identification of specific complementation groups by exploiting the characteristic of FA cells to undergo arrest in the G2/M phase of cell cycle in response to DNA damaging agents. Retroviral vector complementation is currently available for FANCA, FANCC, FANCG, FANCE, FANCF, FANCI, FANCL and FANCB (these complementation groups represent greater than 90% of the FA patients in the US) (FANCM is being developed). A complementation group is identified when correction in the G2/M arrest occurs after transduction with a specific retrovirus vector. Our initial screening will test for FA groups A, C, and G, which represent ~85% of all patients in North America. If no complementation is found in this screening, further testing will occur for FA groups E, F, and L, and subsequently J and B. Including the establishment of cell lines, the complete complementation assay may require up to 6 months. Cell lines established will be deposited in the FA Cell Repository at CCHMC for future research purposes, which requires a patient consent and is provided to any researcher with IRB-approved protocols in a HIPPA-sensitive fashion.

Pricing Details

Service	Unsubsidized cost	Subsidized Charge to clinician/researcher
Fanconi Anemia Complementation: nine complementation groups	\$1072	\$150
Cell Line: EBV transformed lymphocytes	\$180	\$180
Cell Line: Fibroblasts from skin biopsy	\$180	\$180