



FA Complementation and FANCD2 Western Blot

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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 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.

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, FANCL and FANCB (these complementation groups represent greater than 90% of the FA patients in the US). Complementation tests for FANCL is available for Fibroblast samples. A complementation group is identified when correction of the melphalan-induced G2/M arrest occurs after transduction with a specific retrovirus vector.

Pricing Details TTDSL

<u>Service</u>	<u>Current Fee (valid for samples submitted after July1 2009)</u>
Fanconi Anemia Complementation: A,C,G complementation groups	\$ 1250.-
FANCD2 Western Blot	\$ 635.-
Fanconi Anemia Complementation: E,F,L complementation groups (also B complementation if patient is male)	\$ 1250.-
Cell Line: EBV transformed lymphocytes	\$165.-
Cell Line: Fibroblasts from skin biopsy	\$ 383.-

To provide timely and cost efficient service, our initial screening will test for FA groups A, C, and G, which represent ~85% of all patients in North America. If no complementation occurs with groups A, C and G, a FANCD2 western blot is indicated to determine if there is a defect "upstream" or "downstream" of the FANCD2 modification. This assay is also CAP/CLIA compliant and will be performed on request for an additional fee of \$635.-. If the FANCD2 western blot indicates a upstream defect, we recommend complementation testing for groups E, F and L. Male patients will also be tested at this stage for complementation with the X-linked FANCB. Including the establishment of cell lines, the A,C,G complementation assay will require 3 to 5 months.

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