MYCN Gene Amplification by FISH

Indications for Ordering
Prognostic determination in individuals with neuroblastoma or medulloblastoma

Test Description
- Fluorescence in situ hybridization (FISH) analysis using formalin-fixed, paraffin-embedded (FFPE) tissue
- DNA probes
  - MYCN (2p24)
  - CEP2 (control probe)
- 40 cells evaluated from regions of tumor identified on histopathologic review of a matching hematoxylin and eosin stained section

Tests to Consider

Primary test
MYCN (N-MYC) Gene Amplification by FISH 2007227

Related test
Vanillylmandelic Acid (VMA) and Homovanillic Acid (HVA), Urine 0080470
- Initial test for the diagnosis and monitoring of neuroblastoma

Genetics
Gene – MYCN

Test Interpretation

Results
- Amplified – MYCN:CEP2 ≥2.0
  - Predicts poor prognosis
- Nonamplified – MYCN:CEP2 <2.0

Limitations
Results may be compromised if recommended fixation procedures have not been followed

Disease Overview

Incidence – 4th most frequent tumor in children <15 years

Prognostic issues
- Several prognostic factors have been identified in neuroblastoma
  - Individual’s age
  - Stage of disease
  - Marker status
- MYCN is amplified in ~25% of neuroblastomas
  - Associated with poor prognosis, advanced stage, and rapid tumor progression
  - Prognosis depends on level of amplification
  - Prognostication independent from clinical/histopathologic findings
- MYCN is amplified in ~5% of medulloblastomas
  - Associated with clinically aggressive tumors
- MYCN amplifications occur in other tumors, but influence on prognosis is not clear