Fluorescence in-situ Hybridization (FISH)

7q11.23 deletion (Williamssyndrome)

Specimen :

Clinical Indication :

Result :

:

Interpretation :

Total metaphases analyzed	Normal metaphases 2 Orange 2 Green signals	Abnormal metaphases 1 Orange 2 Green signals
20		

PHOTO

Method: FISH analysis performed on 20 metaphases.

Probe: LSI ELN Spectrum orange/LSI D7S486, D7S522 Spectrum green.

Comments: Williams syndrome (WS) is a genetic disorder that occurs in 1/20,000 to 1/50,000 live births. Although WS is typically a sporadic disorder, familial cases have been reported.WS is a contiguous gene deletion syndrome, caused by deletion of several genes on chromosome 7q. One gene that often is deleted in WS is the elastin gene, which causes SVAS and other cardiovascular disease in these patients.The elastin gene, ELN, has been mapped to 7q11.23 (Williams syndrome chromosome region, and is reportedly hemizygous in up to 96% of patients with WS. The deletion of an elastin gene locus cannot be detected by conventional high-resolution chromosome analysis in the vast majority of cases due to the small size of this deletion.