Fluorescence in-situ Hybridization (FISH)

22q11 deletion (DiGeorge syndrome)

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 TUPLE1 (HIRA) (22q11.2) spectrum Orange/LSI ARSA (22q13) spectrum Green.

Comments: DiGeorge syndrome / Velocardiofacial syndrome (VCF)are autosomal dominant conditions with variable expression showing 22q11 deletion. Children typically present with heart defect, craniofacial anomalies and mental retardation. VCF syndrome also show palatal clefting. Most cases are denovo, but about 10% are inherited. In familial cases, a parent can show mild features of the same condition. There is no clear correlation between the extent of the deletion and clinical phenotype.