**ABSTRACT**

A three-year-old male presented with a previous chromosome analysis that revealed an additional unidentified chromosome segment on 13q34, "add(13)(q34)". Metaphase FISH studies initiated utilizing locus specific probes for 13q14 and 13q34 indicated that there was no translocation involving 13q. FISH with a whole chromosome paint probe for 13q confirmed that the extra material was 13q. High resolution chromosome analysis showed a dup(13)(q32.3q33.1). Chromosomal Comparative Genomic Hybridization (cCGH) supported the finding of the FISH assay and helped in an accurate determination of the final karyotype designation as 46,XY,dup(13)(q32.3q33.1).rev ish enh(13q32.3q33.1).

**MATERIALS AND METHODS**

- G band study was done on stimulated pro-metaphase lymphocytes using standard procedures
- FISH was done on interphase and metaphase cells using standard procedures

**cCGH Protocol**

- Extraction of DNA from whole blood
- Direct labeling with FITC (Green label) and Rhodamine (Red label)
- Co-precipitation of control DNA with patient DNA
- Slide Pretreatment with Protease for 5 min

**G-Bands**

- Duplication of bands q32.3 to q33.1 on long arm of chromosome 13 resulting in Trisomy of this region.

**cCGH Results**

- Analysis positive for a duplication of chromosome region 13q32.3 to 13q33.1

**Clinical Features**

- Congenital JET (junctional ectopic tachycardia)
- Pectus carinatum
- Tibial torsion
- Macrocephaly
- Developmental motor delays

**FISH Results**

- LSI 13q34
- LSI D13S319 +13q14
- All probes negative for deletion, duplication or translocation of these regions

- WCP13q
- All material positive for 13q whole chromosome paint

**CONCLUSIONS**

- G-bands identified a duplication
- FISH probes narrowed the region where the duplication was located by detecting no duplications, translocations or deletions above and below the breakpoints identified by G-bands
- cCGH narrowed the region further in conjunction with the above testing to further define and enhance delineation of breakpoints for final interpretation