

Chromosomal Microarray: Comparative Genomic Hybridization CGH-array

Cri Du Chat. *Orphanet J Rare Dis.* 2006; 1: 33. doi: 10.1186/1750-1172-1-33

22q11.2 deletion syndrome. Butler et al. *American journal of medical genetics*, 65(2), 137-141.

1p36 deletion syndrome. Jordan, et al (2015). *The application of clinical genetics*.

Wolff-Hirschhorn syndrome. Sukarova-Angelovska, et al (2014). *Balkan journal of medical genetics*

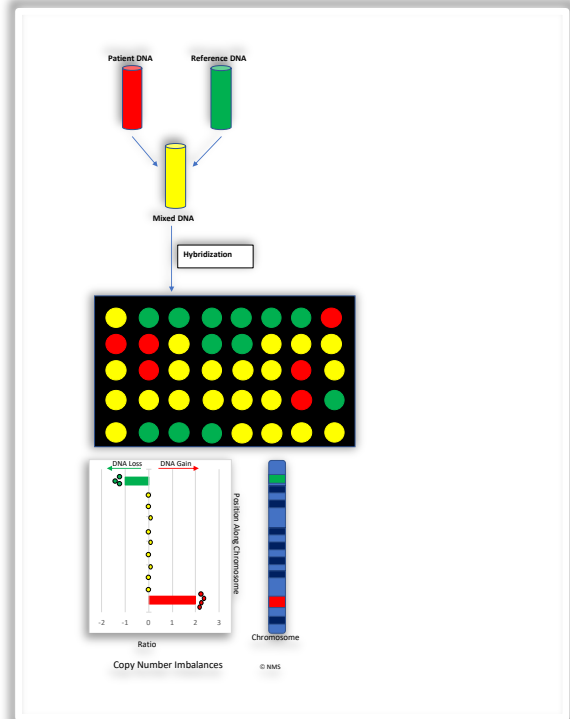


Detects

- Submicroscopic deletions/duplications down to 50-100 kb

Limitations:

- **CAN NOT IDENTIFY:**
 - Balanced Rearrangements
 - Inversions
 - Translocations
 - Point mutations
 - Low level mosaicism
 - Triploidies
 - Uniparental disomy
 - Consanguinity

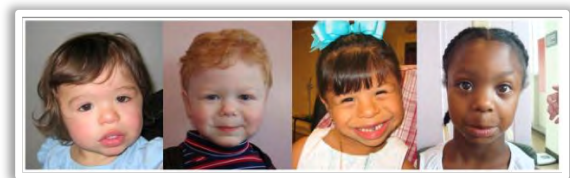


Clinical Applications:

- Multiple Congenital Anomalies
- Developmental delay/ Intellectual disability
- Suspected Microdeletion syndrome

Syndrome	Microdeletion
DiGeorge/Velocardiofacial syndrome	22q11.2
Prader-Willi syndrome/ Angelman syndrome	15q11-q13
Williams Syndrome	7q11.23
Miller-Dieker/Lissencephaly syndrome	17p13.3
Wolff-Hirschhorn syndrome	4p
Cri-du-chat syndrome	5p15.2
ATR-16 syndrome	16p13.3
1p36 Deletion syndrome	1p36

CGH-Array



Young children with Williams syndrome (Left to Right): Asian female, age 19 months; Caucasian male, age 2 years; Hispanic female, age 3 years; African-American female, age 5 years. *Am J Med Genet C Semin Med Genet.* 2010 May 15;154C(2):203-8.