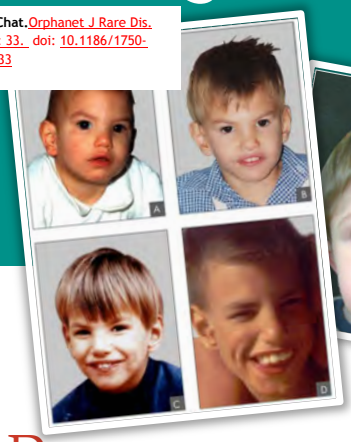
Chromosomal Microarray: Single Nucleotide Polymorphism Array

SNP-array



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

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

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

Wolff-Hischhorn syndrome. Sukarova-Angelovska, etal (2014). *Balkan journal of medical genetics*

Detects

- Submicroscopic deletions/duplications down to 50-100 kb
- Triploidies
- Uniparental disomy
- Consanguinity

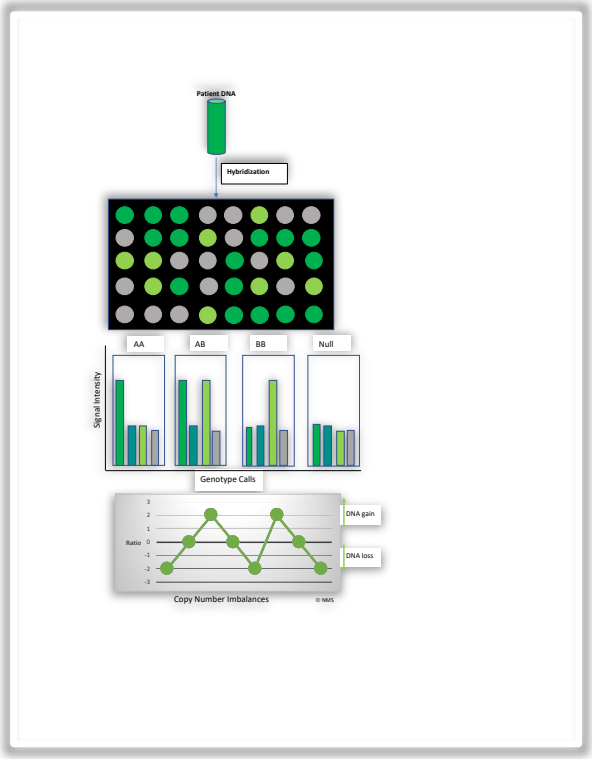
Limitations:

- **CAN NOT IDENTIFY:**
  - Balanced Rearrangements
    - Inversions
    - Translocations
  - Point mutations
  - Low level mosaicism

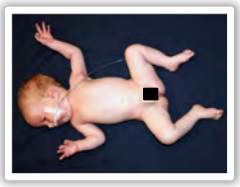
Clinical Applications:

- 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-Hirschorn syndrome	4p
Cri-du-chat syndrome	5p15.2
ATR-16 syndrome	16p13.3
1p36 Deletion syndrome	1p36



SNP-Array



Prader Willi Syndrome

Hypotonia of infancy in a one-month-old male with **PWS**. Note frog leg position and need for a feeding tube. Also note dolichocephaly and hypoplastic, empty scrotum. Eur J Hum Genet. 2009 Jan; 17(1): 3–13.