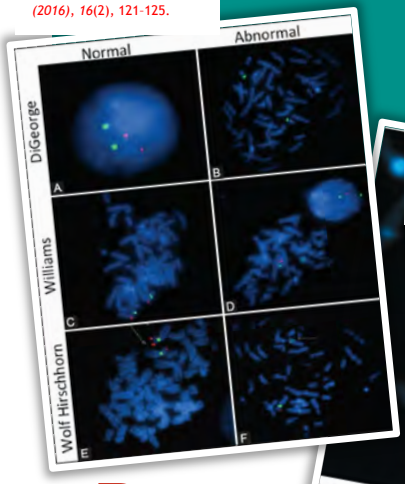
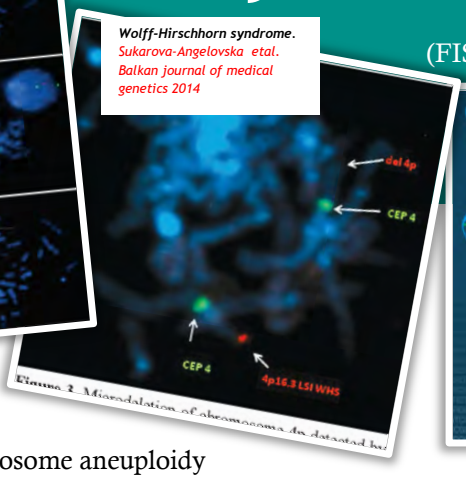
Fluorescence In Situ Hybridization (FISH)



Bosnian journal of basic medical sciences (2016), 16(2), 121-125.



Wolff-Hirschhorn syndrome. Sukarova-Angelovska etal. Balkan journal of medical genetics 2014

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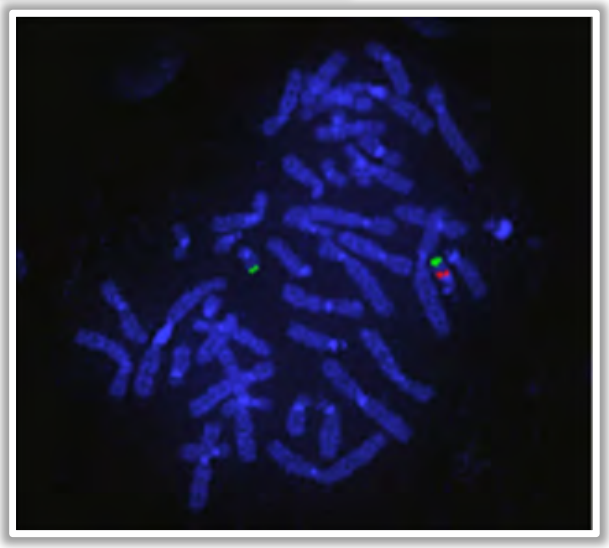
Prader-Willi Syndrome. American journal of medical genetics (1996), 65(2), 137-141.

Detects:

- Whole chromosome aneuploidy
- Microdeletions
- Low Level Mosaicism

Limitations:

- Technical problems related to poor hybridization efficiency
- Only subset of specific aneuploidies and microdeletions are tested (probe dependent)

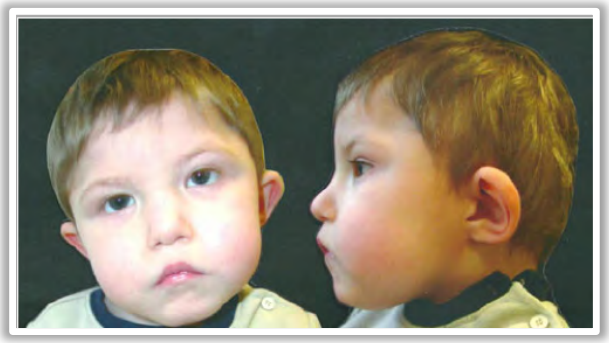


FISH 22q11.2 deletion syndrome. *From Stephane Burtey etal, Kidney International (2008) 74, 1495–1496*

Clinical Applications:

- Karyotyping (rapid)
- Microdeletion Syndromes

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



22q11.2 deletion syndrome. *Butler, M. G., Christian, S. L., Kubota, T., & Ledbetter, D. H. (1996). A 5-year-old white girl with Prader-Willi syndrome and a submicroscopic deletion of chromosome 15q11q13. American journal of medical genetics, 65(2), 137–141.*