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7.62. Supernumerary marker chromosome derived from human chromosome 6: precise definition by inter-species fluorescent *in situ* hybridization with gibbon painting probes

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Small supernumerary marker chromosomes (SMC) are a heterogeneous group of chromosomes with an estimated frequency of approximately 0.14-0.72 per 1000 newborns and higher frequencies in particularly populations such as mentally retarded and infertile males. Routine chromosomal analysis using GTG-banding alone has proved to be limited for the correct identification of marker chromosomes. Even FISH (Fluorescence *In situ* Hybridization) studies with human painting probes are only capable of identifying the origin of the chromosome from which the marker is derived. Microdissection and reverse chromosome painting are being used with success in the precise identification of SMCs and their origin. Here we used an alternative methodology: inter-species *in situ* hybridization with gibbon painting probes for precise chromosomal identification of a supernumerary marker chromosome derived from the human chromosome 6 and to determine its origin.

The SMC(6) was detected by prenatal diagnosis. The GTG analysis revealed a karyotype: 46,XX+mar/46,XX (73,6%/26,4%) in two different cultures. The marker was CBG positive, negative for dystamycin A / DAPI banding and did not contain NOR-positive satellites. FISH identified the chromosomal origin from chromosome 6 with a human painting probe.

The inter-species hybridization with the gibbon probes representative of the chromosomes 1 HCO, 3 HCO, 8 HCO, 17 HCO, 18 HCO and 22 HCO that hybridize to human chromosome 6 allowed the precise identification of the chromosomal region from which the SMC (6) had originated. This alternative methodology, to microdissection and reverse painting, proved to be very useful for the identification of unknown chromosomal structures and the

precise definition of the chromosomal regions present in derivative chromosomes.