

## A rare karyotype presenting monosomy Xp and trisomy 17q in a patient with MCA/DI.

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### Abstract/Resumo

Many chromosome translocations involve microdeletions and/or microduplications leading to chromosomal abnormalities associated with multiple congenital anomalies and mental retardation (MCA/MR). AA, male, children of non-consanguineous and healthy young couple, was born in the sixth month of gestation by cesarean section and gestation without intercurrents, presenting with circular cord delivery, jaundice (phototherapy treatment), remaining in the maternity ward for 6 months in oxygenation. Referred to the SAG for presenting MAC/DI. In the genetic-clinical evaluation at 3 years and 11 months, a history of recurrent pneumonia, cyanosis at birth and to the present, IVC, hypospadias, cryptorchidism, severe psychomotor retardation, speech and physical development. Length of 86cm (<2,5%), weight of 9,700kg (<2,5%), PC of 40cm (<2,0%) e 73cm of wingspan. He presented microcephaly, microstomia, retrognathism, and ears with low implantation. Cytogenetic analysis (GTG and GTG-RA) revealed karyotype 46, XY, t(X;17) (p22.3; q21.2) /46, XY, t(x;17) Xq22.3::17q21.2→17qter,17pter→17q21.2). FISH confirmed the X/17 translocation. Cytogenetic analyzes of parents were normal. A-CGH was revealed partial trisomy 17q and partial monosomy Xp. Microduplication of 17q was 6.5Mb (31004189-37527127) and the monosomy of Xp was 0.9Mb (168551-1102585). This examination also revealed a microdeletion in 17q of 1.2Mb (37532969-38780383). Chromosomal imbalance compatible with the present case was not found in the literature. Approximately 40 cases were found in the literature, of duplication 17q, microdeletion 17q and monosomy Xp. The correlation of phenotype-genotype was found among the genes that are located in the unbalanced regions, which present their impaired functions are: microduplication 17q12-q21.2 - SPACA3, LIG3, RAD51D, GAS2L2, MMP28, LHX1, PCGF2, RPL19 involved with DNA transcription, duplication, repair, embryonic development, tissue remodeling, homeostasis, development of renal, urogenitais systems and skeletal muscle system. 17q12-q21.2 microdeletion are: MED1, NEUROD2, PPP1R1B, TCAP, PGAP3, CSF3, MED24, NR1D1, MSL1, CDC6, GJD3, TOP2A; involved with DNA transcription, neuronal cell differentiation, neurological functioning, regulation of skeletal, cardiac muscle systems, immune system, metabolic, cardiovascular functions and cell division. Xp22.33 microdeletion are: PPP2R3B, SHOX, involved with cellular processes and growth of the individual. The study contributes to support the relationship of the presence of partial trisomy of the long arm of chromosome 17 associated with the presence of a MAC/DI.

Keyword/Palavras-chave: Monosomy Xp; Microduplication; Chromosome 17

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