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UNBALANCED GENOMIC CHANGES AND SMA? : CASE REPORT

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INTRODUCTION

Mental retardation, global developmental delay, epilepsy, autism, neurological syndromes and birth defects can be often linked to rare genetic changes or disorders.

Purpose: To highlight the usefulness of the molecular karyotype.
Keywords: SMA, karyotype, molecular, Array-CGH, disorder

MATERIAL AND METHODS

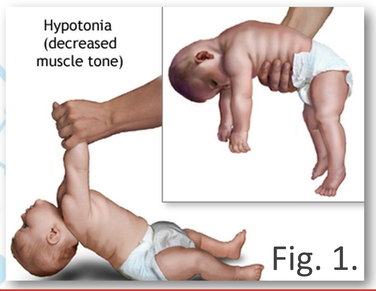


Fig. 1.

We report on a case of 16 months old boy with severe hypotonia(Fig 1), born at term, in a no-consanguineous family. Suggesting SMA, was done the molecular–genetic examination of SMN1 gene through PCR-RFLP method. Considering other clinical manifestations (neuropsychomotor retardation, craniofacial dysmorphia, palmar dermographism, interphalangeal contractures, inverted nipples, hypertrichosis) associated with chromosomal genetic abnormalities, the constitutional karyotype with subsequent molecular karyotype investigation was indicated.

RESULTS

PCR-RFLP result doesn't show deletions of exons 7 and 8. The diagnosis of SMA was excluded. Indication for karyotype was delayed due to the assumption of the presence of a tumor in the cervical region, so the investigation of the Alpha-protein marker had the following results: AFP = 402, ref = 0-23.5, but after 6 days the result was AFP = 9,3, ref = 0-23.5. Elevated AFP results are specific to both a tumor process and a genetic abnormality.

Fig. 2. Chromosome 10. Locus of microdeletion

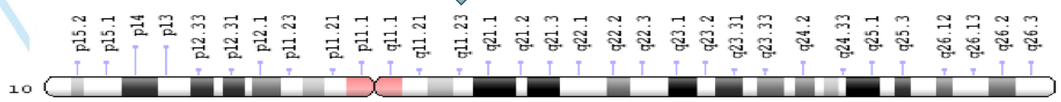
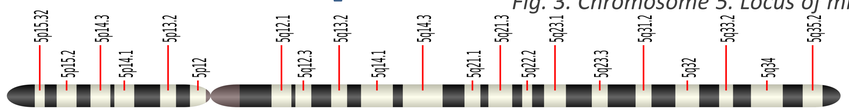


Fig. 3. Chromosome 5. Locus of microdeletion



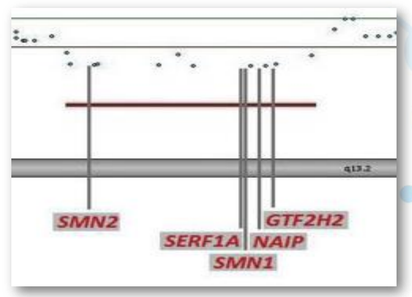
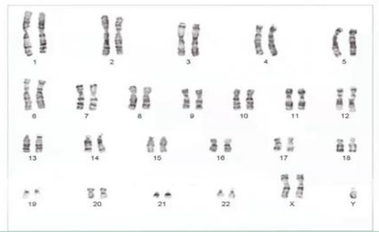


Fig. 4. Genes linked to hypotony and SMA

The result of the constitutional karyotype was normal (46 XY), but the molecular trough Array-CGH method *(fig.5),* identified the following unbalanced haplo-karyotype, insufficient genetic changes: a 1398 Kb microdeletion in the region of chromosome 5q13.2 (of which OMIM registered genes such as SMN1, NAIP, GTF2H2, SERF associated with SMA, *fig.3,4*) and a microdeletion of 4832 Kb in the region of chromosome 10q11.22-q11.23 (of which 6 morbid genes, registered OMIM, *fig. 2*).

Chromosome analysis
Examines a genome *visually*



Chromosome microarray
Examines a genome *comparatively*

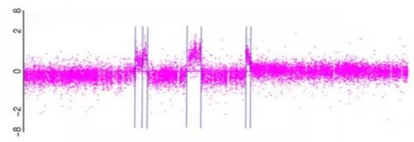


Fig. 5. Karyotype and molecular karyotype difference

CONCLUSIONS.

Molecular karyotype is extremely important in clinical utility for patients with such genomic changes, both in diagnosis and in long-term management and the determination of the risk of recurrence. Its scientific utility is also significant, with new microdeletion or microduplication syndromes being recognized and clinically delineated.

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