

## GESTATIONAL TROPHOBLASTIC DISEASE - THE PATERNAL ORIGIN OF A “MOLE”

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The trophoblastic gestational disease or “*molar*” pregnancy comes from an abnormal growing of the human trophoblastic placenta and also from the male genome. This kind of abnormality originates from a pathology in which the “*moles*” came from a female cell without a nucleus that is fertilized by a male cell (sperm). From this is clear that the genomic charge comes from the father rather from the mother. That means that the origin of chromosomes is masculine, therefore it can't be compared with the mother, who does not contribute to the son's genotype.

During sexual reproduction, the egg and sperm fuse (fertilization) to generate a new individual, i.e. each parent contributes half of their genetic information to generate an individual whose genetic makeup is diploid, so that genetic information provided from both parents give the individual characteristics.

Based on the above, the objective of this study is to determine the paternal origin of the mole.

### Materials and Methods

10 to 12 grapelike vesicles were treated with DTT 1M and Proteinase K (20 mg/ml) in incubation buffer of the Tissue and Hair Extraction kit (Promega, Madison, WI, USA). Prepared lysis solution (100 µl) was then added to the sample and incubated at 56°C for 1 hour. DNA was purified with DNA IQ Kit (Promega, Madison, WI, USA). Purified DNA was collected in 50µl of elution buffer. Blood samples were processed using the FTA card.

### DNA amplification (PCR)

Multiplexed PCR amplification of 15 STR loci, were performed using the Identifiler genomic system (Applied Biosystems). PCR was carried out in a GeneAmp 9700 Applied Biosystems thermal cycler (Foster City, USA).

### DNA fragment analysis

PCR-amplified fragments were analyzed with a Genetic Analyzer ABI PRISM 3130 (Applied Biosystems). Fragments were analyzed based on the allelic ladders contained in each Identifiler kit using GeneMapper Software (version 3.2 (Applied Biosystems)).

### Results

The genotype obtained from the vesicles (moles) show a female homozygous pattern that is not related to the mother's genotype. The analysis from the genetic profile observed for the alleged father shows that for each marker that forms the profile of the mole genotype, there is half its information. This allows us to state that the donor of the genetic information in the molar tissue is the presumed father.

### Conclusion

The results confirm that the genetic profile observed for the mole allows to state that the biological origin of the mole is the alleged father, therefore is not possible to excluding him as the mole biological father.

In the present work we did not show the maternal contribution of the mole. Further work will be directed to study the maternal lineage using the mitochondrial analysis of the mole DNA.

### References

1. Berkowitz, RS, Goldstein DP. Chorionic Tumors. N. Engl. J. Med. 1996,335:1740-1748.
2. Ezpeleta, JM, López, CA. Enfermedad trofoblástica gestacional. Aspectos Clínicos y Morfológicos. REV. ESP. PATOL. 2002, Vol. 35, n° 2: 187-200. ☞