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Familial transmission of translocation 11q;22q

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The constitutional translocation between chromosomes 11 and 22, t(11;22)(q23;q11), is the most frequent chromosomal rearrangement in humans. The risk of having an imbalanced child is greatly increased for the women carrier as compared to men carrier. We found 3 cases with familial translocation t(11q;22q). In these families fetal chromosomal rearrangements were detected by amniocenteses which were ordered because of the advanced matermal age, the abuse of drugs or previous fetal loss. We performed chromosome analysis of parents to identify the origin of fetus' karyotype retrospetively using G-banding. Two fetuses were balanced translocation with 46,XX and XY,t(11;22)(q23;q11)pat. Their fathers were detected later to be a balanced translocation carrier. The remaining fetus was examined as an unbalanced karyotype, 47,XX,+der(22). The derivate chromosome 22 has inherited from mother with balanced translocation 46,XX,t(11;22)(q23;q11).

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Comparison of the Nested PCR Coupled Reverse Transcription with Cytogenetic Analysis for Detection of *BCR/ABL* Rearrangement

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Chronic myelogenous leukemia (CML) is hematological malignancy characterized by excessive growth of myeloid cells and their progenitors. The specific genetic alteration of CML is the formation of the Breakpoint Cluster Region (BCR)/abl fusion gene in leukemic cells. The presence of the BCR/abl gene has important diagnostic and prognostic implications in CML. The detection of BCR/abl gene rearrangement by nested polymerase chain reaction (PCR) coupled reverse transcription was investigated and compared with Philadelphia (Ph) chromosome positive documented by cytogenetic analysis in CML patient samples. In a total of 68 CML patient cases, BCR/abl gene rearrangement were found to have 62 cases (91.2%) by the nested PCR method. On the other hand, 53 cases (77.9%) was detected the Ph chromosome by cytogenetic analysis. The nested PCR products of BCR/abl fusion gene were confirmed by the sequencing analysys. This approach is simple and can routinely be used in most cases on blood and bone marrow cells of patients at the first stages of clinical presentation of CML. From the results, the nested PCR method was able to detect a Ph chromosome in patients in whom the cytogenetics are negative despite a clinical presentation of CML, and can monitor minimal residual disease after bone marrowtransplantation.