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CLINICAL MANIFESTATION OF *DE NOVO* DUPLICATION AT THE DISTAL END OF CHROMOSOME 7P: CASE PRESENTATION

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We report the case of a one and a half month old baby boy with *de novo* duplication at the distal end of chromosome 7p who visited our facility at SSKM Hospital. The clinical manifestations associated with duplication at the end of 7p are mostly delineated and in most cases it results from inheritance of parental chromosome harbouring a balanced translocation. In this study, we describe *de novo* duplication and review the previously published literature from other parts of the world dissecting the genotype-phenotype correlation in such cases. The phenotype for duplication at the distal arm of chromosome 7p is well described. The clinical features associated with such chromosome anomaly comprises of large fontanelles and sutures, hypertelorism, large, apparently low set ears, high arched palate, hip joint dislocation or contractures, a high frequency of cardiac septal defect, and mental retardation. The infant who was brought in our facility was similarly diagnosed of with developmental, cardiovascular, pigmentation problems. However, in most cases such partial trisomy or segmental duplication occurs because of mis-segregation of the chromosome or mis-alignment of the chromosomes and are inherited from parents in form of balanced translocations. In this article, we present the case of possible partial *de novo* 7p duplication depicting the same clinical features and are of utmost importance as it indicates pure segmental imbalances which reflect genotype-phenotype correlations accurately. The distal end around the 7p 21.2 region seems to be important with respect to development and reports on molecular analysis of this region reveal three gene that are very important with respect to development of the embryo viz. *TWIST* , *GLI-3* and *HOX* A. The various reports present indicate that *TWIST* gene in triple dosage at the distal end of chromosome 7 may be causally related to the presence of a large anterior fontanelle with delayed closure, which is the more characteristic clinical feature of the 7p duplication syndrome as also represented in our case study. The presence of cardiovascular problems in our case is unique to other such case representation.

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