# Clinical Impact of Chromosome 16 Short Arm Duplication in the Region 16p12.3p13.11

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The purpose of this research is to indicate the symptoms of a rare duplication located on the chromosome 16 and to propose more efficient way to diagnose the 16p12.3p13.11 duplication syndrome. I analysed the blood sample of patient with hypertrophic pyloric stenosis, neurodevelopmental delay and several other dysmorphic features. The traditional karyotype method was used as well as array- Comparative Genomic Hybridisation. It appeared that patient carries the duplication in the region 16p12.3p13.11. In order to confirm the aberration, fluorescent hybridisation in situ with probes specific to the duplication region was used. After the signal analysis, the mutation was validated. Not finding any information in the professional literature I analysed role of genes mapping in that region. I combined their potential role with the phenotypic manifestation of duplication. I proved that duplication may result in various phenotypic manifestation, particularly in infantile hypertrophic pyloric stenosis. The danger of the disease comes from the dehydration and electrolyte disturbance and may be fatal without early surgery. Research indicated that the aberration may be inherited from healthy parent (it manifests incomplete penetration) who is a carrier. It was confirmed by a probe designed previously for the patient. In that case, when a person-carrier is expecting a child, the prenatal testing may be performed with FISH method and a designed probe and potential surgery may be performed in order to protect the life of a newborn. The knowledge of this phenotypic manifestation results in more accurate predictions which lead to cheaper and quicker diagnostic procedures.

**Awards Won:**