# "A Prenatally Ascertained De Novo Terminal Deletion of Chromosomal Bands 1q43q44 Associated with Multiple Congenital Abnormalities in a Female Fetus"

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## Summary

It is known that the loss, or deletion, of chromosomal material has deleterious effects on the phenotype of the subject affected. This paper involves the study of a deletion of part of chromosome 1, at location 1q43q44, detected prenatally in a 32-year old pregnant woman with abnormal ultrasound findings. It should be noted that the case presented in the current study is the first prenatal case with the smallest, pure 1q43q44 deletion, thus involving no other changes in DNA constitution at this region.

This deletion has been associated with multiple congenital abnormalities; however clinical features may vary amongst patients.

Most patients show characteristic features such as moderate-to-severe intellectual disability, limited to no speech, dysmorphic facial features including round face, prominent forehead, flat nasal bridge, hypertelorism, epicanthal folds, and low set ears. Hypotonia, poor growth, microcephaly, corpus callosum abnormalities (CCA), and seizures are also commonly present in these patients.

Parental karyotypes revealed that the deletion was *de novo*, and additional molecular techniques, MLPA and array-CGH (Comparative Genomic Hybridization), confirmed and 8Mb deletion at this location.

Genetic counseling was offered to the couple and termination of pregnancy was decided at 28 weeks of gestation. The female fetus was sent for autopsy, and further revealed characteristics of microcephaly, microretrognathia, cleft palate, cardiac defect, and small cerebellar vermis.

It is important to highlight the complexity of gene implication in such cases, and the difficulty of genotype-phenotype correlation. Reporting such cases, may contribute to better understanding of these issues.