**Wilkie, Andrew O.M, Zequn Tang, Navaratnam Elanko, et al. "Functional Haploinsufficiency of the Human Homeobox Gene MSX2 Causes Defects in Skull Ossification." 24.Nature Genetics (2000): 387-90. Print.**

The article of Functional haploinsufficiency of the human homeobox gene MSX2 causes defects in skull ossification is a review article. The researchers give an overview of all the research conducted and what their results ended up being. Haploinsufficiency means when diploid organisms only have a single functional copy of a gene. This paper addresses the question if MSX2 dosage is critical for human skull development and if PFM and craniosynostosis result.

The article explains the relationships in multiple families and the MSX2 gene. The researchers explain the other genes and diseases tested for and how the results just didn’t match up. In the first family, researchers concluded that MSX2 was hemizygous in the affected individuals, which would result in PFM (enlarged parietal foramina). In the next 8 sporadic patients or families, the MSX2 gene was again analyzed. To test allele discrimination the intragenic microsatellite and direct sequencing were used. The researchers obtained normal results in six of the eight cases and excluded linkage to MSX2 in two of them. Those two families confirmed that MSX2 is a disease for non-syndromic parietal foramina. The researchers then concluded that the likely mechanism of these MSX2 mutations is haploinsufficiency.

The data in the paper was stated, only some was shown. For the eight patients and families, PAC contig (filled bars) was shown showing the positions of exons 1 and 2 of MSX2 in relation to the intragenic. The data shows where the mutated gene sequence breaks to result in the MSX2 gene malfunction.

The quality of the evidence in this paper is high. There were nine people who worked on the experiments and this review article. The researchers worked from all different hospitals around the world. Andrew O.M. Wilkie was from Institute of Molecular Medicine, Department of Clinical Genetics and the Craniofacial unit ad Radcliffe. The researchers ranged from the UK to the USA to Poland.

The conclusions are important, because they are leading to find out what MSX2 results in and whether or not this is critical for human skull development. There are only few children around the world, including my brother who are missing this gene. This gene holds entail for them the rest of their life. This research has given some more insight to doctors to find out what is wrong with them. The missing skull bone can lead to more problems, as such in my brother, including brain and eye hemorrhaging. These children, since the skull is not fully developed cannot play sports. If the doctors learn more about this gene, they may be able to find a way to increase the bone growth. .