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The Genetic Basis Of Patent Foramen Ovale

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Background: Patent foramen ovale (PFO) is a remnant of normal fetal development that persists in ~25% of the population. It is a canal connecting the atria that allows the transit of blood from venous to arterial circulation, bypassing the pulmonary circuit. PFO is associated with cryptogenic stroke, migraine, visual auras, and other medical conditions. Based on the analysis of family pedigrees, previous studies suggest that PFO is an inherited condition. The aim of this novel study was to elucidate the specific genes involved in maintaining patency of the foramen ovale after birth.

Methods: Medical records of patients with identified PFO were reviewed for family history of PFO prevalence. Of 750 patients with PFO, 26 families were identified having multiple members diagnosed with PFO. Five families (16 individuals) were recruited for genetic testing. PFO was diagnosed using transcranial Doppler imaging with agitated saline. Serum DNA was collected from the 16 subjects, and was analyzed using exome sequencing. A two-tailed t-test analysis was used to compare the mutations between the groups of those with PFO and those without PFO; p value < 0.001 was considered as statistically significant.

Results: In the five families (16 individuals) with available DNA samples, PFO was found in 10 patients (62.5%). Exome sequencing revealed 25 mutations on 13 unique chromosomes that demonstrated statistically significant differences between the two groups of those with PFO and those without PFO ($p=0.0002$).

Conclusions: Genetic sequencing provides specific mutations that are linked to patency of the foramen ovale. Further genetic testing on more families is ongoing.