Imaging Findings in a Familiar Case of Foramina Parietalia Permagna

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Purpose
Parietal foramina has been found to be caused by mutations in the MSX2 gene. The aim of this study is to present a three generation family with spectral enlarged parietal foramina and other associated brain anomalies.

Materials & Methods
Three patients of the same family (son, mother, and grandfather) were evaluated by genetic and imaging studies. The imaging modalities used were X-ray of skull (PA and lateral), CT without contrast, 3D CT bone reformatted images and also MR imaging and MR venography, used to identify brain anomalies associated with this considered benign calvarial defect.

Results
The imaging studies of all three patients showed distinct variations and expressions of the same following alterations: calvarial defect consistent with foramina parietalia permagna, persistent falcine venous sinus, deep white matter signal changes, high insertion of the tentorium with enlarged posterior fossa and some variations of the paramedian occipital infolding.

Conclusion
Foramina parietalia permagna is an MSX2 gene defect expressing in the imaging studies with morphostructural changes in the brain involving not only the calvarium but the cortical infolding, white matter, and cerebrovascular drainage system.

References
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