Letter to the Editor

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Radiomic Models for Diagnosing Juvenile Myoclonic Epilepsy Should Note Its Genetic Heterogeneity

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I read with interest the article by Kim et al. [1] about the generation of magnetic resonance imaging (MRI)-derived radiomic models to diagnose juvenile myoclonic epilepsy (JME). Seven radiomic models (light gradient boosting machine, support vector classifier, random forest, logistic regression, extreme gradient boosting, gradient boosting machine, and decision tree) were examined [1]. The light gradient boosting machine achieved the highest area under the receiver operating curve, without significant differences with other models [1]. The putamen and ventral diencephalon were the most important for suggesting JME [1]. MRI-derived radiomic models differentiated JME from healthy controls [1]. The study is appealing but needs further discussion.

The main study limitation is that JME is considered as a single entity, although there is evidence that this is not the case [1]. JME can be idiopathic or hereditary [2]. Hereditarily, mutations in several genes, such as *CACNB4*, *EFHC1*, *NKCC1* (*SCL12A2*), *KCC2* (*SCL12A5*), *CSTB*, and *GABRA1* [2,3], were reportedly associated with JME [2]. The genetic heterogeneity of the disease suggests that cerebral functions may be differentially impaired in these JME subtypes. Additionally, there is evidence that the methylation status of certain DNA segments is associated with JME expression [4].

Current antiseizure drug (ASD) treatment was not considered in the evaluation. ASDs can strongly influence the clinical and subclinical seizure activity extent and therefore, the models generated [1]. Particularly, the amount of epileptiform discharges and background activity can strongly determine the predictive value of the applied models.

Of the 97 patients, the number of patients that have and did not have structural abnormalities on cerebral MRI was not reported. Structural abnormalities can strongly define a radiomic model. Generally, patients with JME can be divided into JME with and without morphological abnormalities on cerebral imaging [2].

Patients with JME were only compared to healthy controls, but not to other types of epilepsy [1]. Knowing whether the models used can also differentiate JME from other types of epilepsy, it is crucial to assess whether these models can aid in the diagnosis of JME.

Overall, the study has obvious limitations that require re-evaluation and discussion. Clarifying the weaknesses would strengthen the conclusions and improve the study. Before using radiomic models to diagnose JME, the etiology of JME must be clarified. If JME is considered as an independent entity but not as a syndrome with similar clinical and electroencephalographic features and heterogeneous genetic etiology, the diagnosis of the models remains unproven.

Conflicts of Interest

The author has no potential conflicts of interest to disclose.

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REFERENCES

1. Kim KM, Hwang H, Sohn B, Park K, Han K, Ahn SS, et al. Development and validation of MRI-based radiomics models for diagnosing juvenile myoclonic epilepsy. *Korean J Radiol* 2022;23:1281-1289
2. Amrutkar C, Riel-Romero RM. *Juvenile myoclonic epilepsy*. [Updated 2022 Aug 8]. In: *StatPearls* [Internet]. Treasure Island (FL): StatPearls Publishing; 2022 Jan [cited 2022 Dec 11]. Available from: https://www.ncbi.nlm.nih.gov/books/NBK537109/
3. Berrechid AG, Bendjebara M, Bouteiller D, Nasri A, Peuvion JN, Marie Y, et al. Juvenile myoclonic epilepsy phenotype in a family with Unverricht-Lundborg disease. *Epileptic Disord* 2019;21:359-365
4. Pathak S, Miller J, Morris EC, Stewart WCL, Greenberg DA. DNA methylation of the BRD2 promoter is associated with juvenile myoclonic epilepsy in Caucasians. *Epilepsia* 2018;59:1011-1019

 https://doi.org/10.3348/kjr.2022.0982