Alisher J. Yuldashev, Chang Ho Shin, Yong Sung Kim, Woo Young Jang, Moon Seok Park, Jong Hee Chae, Won Joon Yoo, In Ho Choi, Ok Hwa Kim, Tae-Joon Cho, Reply:

We thank Dr. Viana and colleagues for the interest in our study. It was interesting to know about four Brazilian patients with Camurati-Engelmann disease (CED). Making a timely and correct diagnosis is still important to avoid unnecessary diagnostic research and facilitate patients' comfort in daily life. Described clinical manifestations of all four patients can prove phenotypic variability of this rare genetic disease. Among all clinical presentations, pain was the most frequent symptom[1] resulting in resemblance among patients. However, onset of pain and clinical presentation are unpredictable.

The patient described by Dr. Viana and colleagues had clinical symptoms from early childhood. Confirmed hearing loss by audiometry gives information about spreading pathologic process through cranium bones. All signs show active progression of the disease. A genetic test result which confirms mutation of TGFB1 would play a crucial role in making a final diagnosis.

In our paper, we presented clinical manifestations of eight CED patients and classified them according to the mode of initial presentation. As a criterion of classification, the age of onset of clinical symptoms was not taken as a key point. All patients had pain and the severity of pain ranged from intermittent after physical exercise to severe. Absence of straight genotypic and phenotypic correlation[1] makes it difficult to make a timely diagnosis of CED.

Regarding the Brazilian boy suspected to have CED, we appreciate your efforts on investigating and diagnosing as rapidly as possible, although we could not confirm that the patient had CED from the data presented. We express our gratitude for mentioning the CED classification scheme and requesting our opinion. We would like to include him into group II (patients who mainly present with pain) because the pain was the first clinical presentation regardless of the age of onset.

CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

REFERENCE

1. Janssens K, Vanhoenacker F, Bonduelle M, et al. Camurati-Engelmann disease: review of the clinical, radiological, and molecular data of 24 families and implications for diagnosis and treatment. J Med Genet. 2006;43(1):1-11.

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