Letter to the Editor

Reply to Dr Mussa A *et al.*

Dear Sir;

We thank Dr Mussa, Alessandro *et al.* [1] for their comment on our recent article in Endocrine Journal [2]. In this letter, they suggested that our patient could be included as a disorder-related PHACES syndrome. As they pointed out, posterior fossa malformation, hemangioma, arterial anomalies, coarctation of the aorta, and cardiac defects and eye anomalies are all hallmarks of PHACES syndrome [3]. In PHACES syndrome, some patients have also been reported to have congenital hypothyroidism. The patient with PHACES syndrome described in their letter showed congenital hypothyroidism caused by functional defect, ventricular septal defect, and optic atrophy, but no brain anomalies including hemangioma. Thus, PHACES syndrome may have broad clinical phenotypes more than expected.

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Our patient showed congenital hypothyroidism due to functional defect and cerebella atrophy, but no hemangioma, cardiac defect, or optic atrophy. Thus, in the case of our patient, it could not be conclusively determined whether or not he was an atypical PHACES syndrome. As suggested in this letter, clarification of the molecular basis of PHACES syndrome is needed to provide a definitive answer.

References

1. Mussa A, Corrias A, Baldassarre G, Biamino E, Silengo M (2007) Congenital hypothyroidism., cerebellar atrophy, and the incomplete phenotypic expression of PHACES syndrome. Endocr J (Letter).
2. Tajima T, Fujiwara F, Sudo A, Saito S, Fujieda K (2007) A Japanese patient of congenital hypothyroidism with cerebellar atrophy. Endocr J. Epub ahead of print.
3. Bronzetti G, Giardini A, Patrizi A, Prandstraller D, Donti A, Formigari R, Bonvicini M, Picchio FM. (2004) Ipsilateral hemangioma and aortic arch anomalies in posterior fossa malformations, hemangiomas, arterial anomalies, coarctation of the aorta, and cardiac defects and eye abnormalities (PHACE) anomaly: report and review. Pediatrics. 2004 113: 412–415. Review.