Dear Editor,

A 5-year-old boy was referred to our clinic because of pigmentation involving the hands and feet, which had been present since he was 7–8 months old. There was no history of sun sensitivity. He had undergone dilatation operation three times because of achalasia. His parents and relatives were found to be free of similar symptoms. Dermatological examination revealed areas of mottled hyperpigmentation and hypopigmentation involving the dorsa of the hands and feet (Fig. 1a). Mild symptoms of the disease were also found on the knees and there were hyperpigmented freckle-like (1–2 mm) macules on the cheeks (Fig. 1b). Routine laboratory tests were unremarkable. Histopathological examination of skin biopsy specimen taken from a pigmented macule revealed melanin incontinence in the basal layer and a hypopigmented macule was non-specific. The ADAR1 gene was sequenced for our patient and his family members. It detected a pathological mutation, c.1110-1111delCAp.N370fsX373 in exon 2, in our patient and his father (Fig. 2). However, the same mutation was not detected in his mother or sister. Based on these findings, our patient was diagnosed as having dyschromatosis symmetrica hereditaria (DSH).

Dyschromatosis symmetrica hereditaria is an autosomal dominant pigmentary disorder characterized by a combination of hyperpigmented and hypopigmented macules distributed on the dorsal aspects of the extremities and freckle-like macules on the face.1 Although DSH is a rare autosomal dominant genodermatosis predominantly occurring among Japanese and Korean individuals, it occurs in families of every ethnic origin all over the world.1,2 The widest series on this topic was reported by Oyama et al.2 in 1999. In that paper, 185 cases of DSH are reviewed and clinical, histological and genetic features of this condition are delineated. Of the 185 cases, 170 (92%) were reported from Japan.2 To our knowledge, Aliagaoglu et al.3 reported the first Turkish patient that presented hyperpigmented and hypopigmented macules in a generalized distribution with dyschromatosis universalis hereditaria in 2008.3

The cause and pathogenesis of the disease have not been clarified. The pattern inheritance was basically autosomal dominant. On the other hand, there was no family history of DSH in the great majority of cases (114/185) and the autosomal dominant form of DSH is due to a mutation in the DSRAD gene (ADAR1) which encodes a dsRNA-specific adenosine deaminase on an RNA editing enzyme.1,2 Our patient’s parents and relatives were found to be free of symptoms except his father. The novel pathological mutation named c.1110-1111delCAp.N370fsX373 in exon 2 was detected in our case as well as his father. So, we examined his parents and relatives were found to be free of similar symptoms. Dermatological examination revealed areas of mottled hyperpigmentation and hypopigmentation involving the dorsa of the hands and feet (Fig. 1a). Mild symptoms of the disease were also found on the knees and there were hyperpigmented freckle-like (1–2 mm) macules on the cheeks (Fig. 1b). Routine laboratory tests were unremarkable. Histopathological examination of skin biopsy specimen taken from a pigmented macule revealed melanin incontinence in the basal layer and a hypopigmented macule was non-specific. The ADAR1 gene was sequenced for our patient and his family members. It detected a pathological mutation, c.1110-1111delCAp.N370fsX373 in exon 2, in our patient and his father (Fig. 2). However, the same mutation was not detected in his mother or sister. Based on these findings, our patient was diagnosed as having dyschromatosis symmetrica hereditaria (DSH).

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father in detail but there was no finding except a few slightly hyperpigmented macules on the extensor surfaces of his feet, although he described to us that he had had the same hyperpigmented macules on the hands and feet during his childhood but that they had disappeared over time. However, none of the previous studies reported seasonal changes or spontaneous regression with age.

In the great majority of cases, DSH is present as an isolated entity. Our patient was associated with achalasia and this is the first reported case of DSH and achalasia in the same patient. Achalasia is a rare esophageal motility disorder characterized by absent peristalsis and failure of the lower esophageal sphincter to relax. There is increasing evidence that genetic alterations might play an important but underestimated role. This association is probably coincidental but if the two entities are clarified, a significant association may be described between the two facts. We believe that more genetic studies on DSH and achalasia are needed to clarify this association.

ACKNOWLEDGMENT

The authors would like to thank Dr Emel Ergül in Kocaeli University Faculty of Medicine for support in genetic evaluation.

REFERENCES