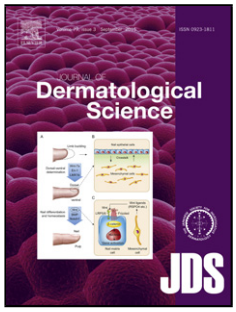

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Clinical application of next-generation sequencing for the diagnosis of segmental neurofibromatosis

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Highlights

- Segmental neurofibromatosis is a rare variant of neurofibromatosis.
- Sanger sequencing from the blood sample could not detect pathogenic mutations.
- NGS could identify a low-frequency pathogenic mutation from the tumor tissue.
- This study revealed the efficacy of NGS in rare disease diagnosis.

Keywords: segmental neurofibromatosis; NF1; next-generation sequencing; somatic mosaicism;

Neurofibromatosis type 1 (NF1) is a complex disorder affecting approximately 1 in 3,500 people [1]. The predominant manifestations include café-au-lait spots on the skin, Lisch nodules, and neurofibromas affecting any region of the body. There are several subtypes of neurofibromatosis based on clinical diagnostic criteria established in 1987 [2]. Segmental neurofibromatosis (NF5) is a rare form of neurofibromatosis defined by regionally limited cutaneous signs of NF1 [3-5]. It is generally thought to result from a postzygotic *NF1* mutation, leading to somatic mosaicism. Thus, it is also called “mosaic localized NF1” [6, 7]. Such mosaicism explains the various clinical manifestations of segmental neurofibromatosis [8]. The phenotypes are known to be closely associated with the