Abstract

Deactivating germline mutations in LEMD3 cause osteopoikilosis and Buschke-Ollendorff syndrome, but not melorheostosis.

Introduction

Melorheostosis (MEL; OMIM #155950) is a troublesome, not well-characterized skeletal dysplasia characterized by “melting” bones of the extremities occurring after puberty. The project aims to study patients with melorheostosis in order to better understand this disease.

Melorheostosis (MEL) is a benign, unstable skeletal dysplasia characterized by “melting” of bones in the extremities occurring after puberty. The project aims to study patients with melorheostosis in order to better understand this disease.

Methods

We investigated 11 patients with melorheostosis. Ten patients were diagnosed as having melorheostosis based on clinical and radiographic findings. Two patients were diagnosed as having osteopoikilosis and melorheostosis based on clinical and radiographic findings.

Results

LEMD3 analyses were performed in 10 patients with melorheostosis and osteopoikilosis. The results showed that deactivating germline mutations in LEMD3 cause osteopoikilosis and Buschke-Ollendorff syndrome, but not melorheostosis.

Discussion

Deactivating germline mutations in LEMD3 cause osteopoikilosis and Buschke-Ollendorff syndrome, but not melorheostosis.

References


Conclusion

Deactivating germline mutations in LEMD3 do not cause classic sporadic melorheostosis.