

Nailing down the diagnosis: an unusual cause of Paronychia.

Dr. Daniely

Paronychia is a common diagnosis, with various infectious and environmental etiologies. I will present a unique clinical case of a family with recurrent episodes of toenails paronychia since childhood, along with facial dysmorphism, alopecia, eyelashes trichomegaly and short stature, that was diagnosed in our outpatient clinic with Costello syndrome. Whole exome sequencing revealed a frame shift mutation in the gene H-ras, causing transcription alterations of the two isoforms of H-ras- P19 and P21, as previously described by Pantaleoni et al. Costello syndrome is a rare Rasopathy with major advances in the understanding of its pathomechanism in the last two decades, usually caused by a gain of function missense mutation. However, in light of the variable genetic changes recently described, it seems that Costello syndrome represent a clinical spectrum with variable phenotype presentation.