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## Telomerase mutations

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Patients with **dyskeratosis congenita** suffer from defects in skin, nails, hair and gut, and usually die from bone-marrow failure. An X-linked form of the disease is due to mutations in the **DKC1 gene** that encodes dyskerin, a protein associated with small nucleolar RNAs and telomerase RNA (hTR). In the September 27 **Nature**, Tom Vulliamy and researchers at **Imperial College** and the Hammersmith Hospital, London, UK, identify mutations that cause autosomal dominant dyskeratosis congenita (*Nature* 2001, **413**:432-435). A genome-wide scan using 400 microsatellite markers narrowed down the search to chromosome 3q21-3q28 which includes the hTR gene. They found an 821 bp deletion including 74 bp 3' of the coding region, and point mutations in some dyskeratosis congenita families. The authors suggest that haplo-insufficiency of hTR accounts for telomere erosion and **premature-aging** symptoms associated with the dyskeratosis congenita disease.

## References

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