MUTATED SPRTN CAUSES:

1. progeroid features
&
2. early-onset hepatocellular carcinoma

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SPARTAN / DVC1 / C1orf124
- Y2H (Ub) screen
- Bioinformatic screen for UBZ4

(Bienko M et.al., Science 2005)
Werner like progeroid syndrome

Family A
Mutations in *SPRTN* cause early onset hepatocellular carcinoma, genomic instability and progeroid features

The figure shows that mutations in *SPRTN* lead to increased UV-induced mutagenesis, which results in genomic instability, early-onset HCC, and premature aging. The protein SPRTN is involved in translesional DNA synthesis and the prevention of replication stress.
Conclusions:

- DVC1 dysfunction leads to progeroid syndrome
- Bialelic DVC1 mutations results in susceptibility to HCC