Condition: Progeria (Hutchinson-Gilford progeria syndrome)

Inheritance:
de novo dominant mutation.

Genetic etiology:
Mutation in *LMNA* gene that encodes lamin A/C protein.

Frequency:
Approximately 1/4,000,000 births.

Clinical features:
Though normal at birth, failure to thrive occurs in early childhood, with development of characteristic facial appearance including loss of hair and subcutaneous fat. Severe atherosclerosis develops, which is the usual cause of death. Intelligence is normal.

Management:
Supportive care; clinical trials underway with farnesyl transferase inhibitors.

Genetic counseling:
Cases almost always sporadic, with rare possibility of germline mosaicism.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.