Opis choroby *

Definicja

A rare, severe, circulatory system disease characterized by premature, diffuse, severe atherosclerosis (including the aorta and renal, coronary, and cerebral arteries), sensorineural deafness, diabetes mellitus, progressive neurological deterioration with cerebellar symptoms and photomyoclonic seizures, and progressive nephropathy. Partial deficiency of mitochondrial complexes III and IV in the kidney and fibroblasts (but not in muscle) may be associated. There have been no further descriptions in the literature since 1994.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Atherosclerosis-hearing loss-diabetes-epilepsy-nephropathy syndrome
Zespół Feigenbauma, Bergerona i Richardsona
Feigenbaum-Bergeron-Richardson syndrome

Kod ORPHA
1192

Kod OMIM
209010

Kod ICD10
I70.9

Kod ICD11
LD2H.Y

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*Źródło

orphanet