

Mięsak klatki piersiowej z deficytem SMARCA4

Kod Orpha: 466962 Kod OMIM:

Opis choroby *

Definicja

A rare soft tissue tumor characterized by a compressive mass located in the mediastinum and/or pleura and lung, including prominent lymph node involvement, histologically poorly differentiated and frequently showing rhabdoid features. Loss of SMARCA4 is typically accompanied by SMARCA2-deficiency. Presenting symptoms include dyspnea, cough, chest pain, or dysphagia, among others. The tumors are aggressive with limited response to chemotherapies, rapid local progression, high recurrence rate after surgical resection, and short median survival times. There is a strong association with smoking.

Dane

Klasyfikacja

Choroba

Synonimy

SMARCA4-deficient thoracic sarcoma

SMARCA4-deficient thoracic sarcoma

Kod ORPHA

466962

Kod OMIM

-

Kod ICD10

C49.3

Kod ICD11

-

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.