



Internal Medicine Flashcard

Extra- and intra-pulmonary nodules

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1. Indication

A 43-year-old female with neurofibromatosis type 1 (NF1) presented with gradual onset of shortness of breath and back pain for several months. She also noticed swelling over the right aspect of her anterior chest wall (Fig. 1a). She had no history of fever, cough, sputum, trauma, or weight-bearing. Physical examination revealed multiple nodules and two larger masses over the trunk, decreased right breath sounds, and dullness on percussion. Computed tomography (CT) findings are shown in Fig. 1b and c.

What is the diagnosis?

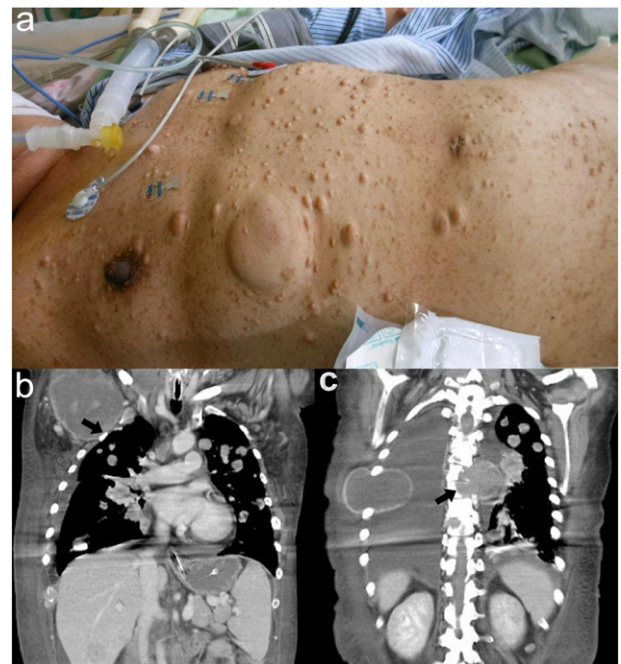


Fig. 1. (a) multiple nodules over the trunk and a protruding mass from the chest wall. (b, c) computed tomography reveals multiple lung nodules, pleural effusion, and tumor invasion to the spine and ribs (arrows).

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2. Diagnosis

CT revealed multiple lung nodules, pleural effusion, and tumor invasion to the spine and ribs (arrows, Fig. 1b and c). CT-guided biopsy provided a diagnosis of malignant peripheral nerve sheath tumor (MPNST). After the diagnosis the patient underwent palliative care.

NF1 is an autosomal dominant syndrome resulting from a diverse range of mutations in the *NF1* gene. Individuals with NF1 have an approximately 10% lifetime risk of developing MPNST, which most commonly occurs in the third or fourth decade of life [1]. In contrast, MPNST in the context of NF1 occurs in approximately 50% of cases; the remainder cases arise sporadically or following radiation therapy [2]. In this report, skin (extra-pulmonary) nodules indicated typical NF1 features and intra-pulmonary nodules indicated metastasis of MPNST. There is a high rate of relapse following multimodality therapy in early disease and a low response rate to cytotoxic chemotherapy at the advanced stage. Prognosis is generally poor, with an overall 5-year survival rate of approximately 33% [3].

Conflict of interests

There are no competing interests.

Contributorship

Ya-Mei Tsai and Pin-Zuo Ko wrote the report.

Ya-Mei Tsai and Te-Chun Shen provided case details and planned the report.

Te-Chun Shen supervised the report.

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