Osteosarcoma in a Boy with Rothmund-Thomson Syndrome

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Abstract. The first case of osteosarcoma in a 13 year old boy with Rothmund-Thomson syndrome is reported. The association between these two conditions is uncertain.

Key words: Rothmund-Thomson syndrome – Osteosarcoma

The purpose of this paper is to report a case of osteosarcoma in a boy with Rothmund-Thomson syndrome (Fig. 1).

Case Report

This 13 y. o. boy was admitted to the Royal Alexandra Hospital for Children, Sydney on 30.12.1977 with a 6 month history of

Fig. 1. Boy 5 y. o. Rothmund-Thomson syndrome. Note skin changes

Fig. 2. 2 y. o. There is some hypotubulation of the forearm bones with a slightly distorted trabecular pattern. Aplasia of the thumb. Irregular hypoplasia of the phalanges

Fig. 3. 6 y. o. Abnormal trabecular pattern. The metaphyses and epiphyses of the knee are most severely affected. Note cyst-like defects in the distal r. femoral metaphysis

Fig. 4 a and b. 11 y. o. L. knee. Abnormal trabecular pattern. Note abnormal ossification of the patella and multiple lines of arrested growth

Fig. 5. 13 y. o. Abnormal trabecular pattern. Typical changes of osteosarcoma at the proximal end of the shaft of the l. tibia