The Expanding Family of Ewings and related tumours

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The Expanding Family of Ewings and related tumours
Ewings sarcoma, EWSR1-Fli1 fused
The Expanding Family of Ewing’s and related tumours

"...it has recently been appreciated that there are cases of Ewing-like sarcomas [...] that lack rearrangement of EWSR1 or other TET family members [...], such as t(4;19) [...] encoding a CIC-DUX4 fusion. Whether these cases should be defined as Ewing sarcoma, or represent separate tumour types is unknown, but currently they are treated in a similar fashion. For the subgroup of “Ewing-like sarcomas” characterized by the BCOR-CCNB3 fusion, a distinct gene-expression profile [...] provides strong arguments for regarding it as a separate entity [...]."

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BCOR-CCNB3 (Ewings like) sarcoma

A new subtype of bone sarcoma defined by BCOR-CCNB3 gene fusion

Gaëlle Pierron¹,⁴, Franck Tirode²,⁴, Carlo Lucchesi²,⁴, Stéphanie Reynaud¹, Stelly Ballet¹, Sarah Cohen-Gogo², Virginie Perrin², Jean-Michel Coindre³ & Olivier Delattre¹,²
from Pierron et al. Nat Genet 2012
BCOR-CCNB3 (Ewings like) sarcoma

RT-PCR screening of 594 sarcomas
(EWSR1-ETS, EWSR1-NFATC2, EWSR1-WT1, SYT-SSX, PAX3-FOXO1, PAX7-FOXO1 negative)

→ 24 BCOR-CCNB3 fusion sarcomas, 19 in bone, all in adolescents/young adults

from Pierron et al. Nat Genet 2012
BCOR-CCNB3 (Ewings like) sarcoma

ROH Birmingham: screening of 134 monotonous high grade small cell fusion negative sarcomas with CCNB3 immunohistochemistry.

→ 10 CCNB3 positive sarcomas, 9 confirmed by RT-PCR
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