Solid Tumour Section
Short Communication

Soft tissue tumors: Desmoplastic small round cell tumor

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Published in Atlas Database: May 1999
Online updated version : http://AtlasGeneticsOncology.org/Tumors/desmoplasticID5023.html
DOI: 10.4267/2042/37544
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Identity
Alias
\( t(11;22)(p13;q11) \) in desmoplastic small round cell tumor

Clinics and pathology

Phenotype / cell stem origin
Thought to be of peritoneal blastomatous cell origin.

Epidemiology
Rare; mostly in childhood and adolescent males (mean age: 22 years; sex ratio: 4.7 M/1F).

Clinics
Very aggressive tumor, located almost exclusively to the peritoneal surfaces of the abdomen with involvement of many abdominal organs; very rare localisations outside the abdominal cavity (thoracic, cranial or skeletal localisations).

Pathology
Characterized by nested pattern of small poorly differentiated tumor cell growth surrounded by dense desmoplastic stroma and immunohistochemical trilineage coexpression: epithelial (cytokeratin, EMA), mesenchymatous (desmin, vimentin) and neural (NSE).

Treatment
Surgery may be performed before intensive chemo and radiotherapy.

Prognosis
Very poor; 35% overall progression-free survival at 5 years; median survival of about 17 months, although tumors are responsive to aggressive therapy in some cases.

Cytogenetics
Note
Besides the specific \( t(11;22)(p13;q12) \), 2 variant translocations have been described.

Additional anomalies
Frequent additional abnormalities, sometimes complex.

Variants
The variant translocations are:
\( t(2;21;22)(p23;q22;q13) \) and \( t(11;17)(p13;q11.2) \).

Genes involved and proteins

EWSR1
Location
22q12
DNA / RNA
Spans over 40 kb, 17 exons; 2.4 kb mRNA.
Protein
656 amino acids; N-term gln-thr-pro-rich region; C-terminal proline rich region; wide expression; RNA binding protein.

WT1
Location
11p13
DNA / RNA
Spans over 50 kb, 10 exons; alternative splicings (in particular, the second site adds or remove 3 amino-acids (KTS) between the second and third zinc fingers and generates 2 isoforms); mRNA 3.5 kb.
Protein
52-54 Kda; 4 Cys2-His2 zinc fingers, glutamine-proline-glycine-rich transcriptional regulation domain, interacting with p53; nuclear localisation; transcriptional repressor.

Result of the chromosomal anomaly

Hybrid Gene
Description
5' EWS - 3' WT1

Transcript
mRNA detectable by RT-PCR (99% of DSRCT).

Fusion Protein
Description
The N-term trans activation domain of EWS (exon 7) is fused to the C-term zinc fingers domain of WT1(exon 8); molecular variants have been described (exons 9 or 10 of EWS); transcriptional activator; the 2 isoforms EWS-WT1 without KTS and EWS-WT1 with KTS having different properties.

Expression / Localisation
Nuclear.

Oncogenesis
Early: in the embryonal mesenchyme of the coelomic cavities; due to inappropriate transcriptional activation of WT1- responsive genes; the EWS-WT1 without KTS isoform would be dominantly acting as an oncogene.

References


This article should be referenced as such: