Atlas of Genetics and Cytogenetics in Oncology and Haematology

OPEN ACCESS JOURNAL AT INIST-CNRS

Solid Tumour Section

Short Communication

t(11;22)(q24;q12) in rhabdomyosarcomas (RMS)

Jean-Loup Huret

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

Published in Atlas Database: June 2010

Online updated version : http://AtlasGeneticsOncology.org/Tumors/t1122q24q12RhabdoID6285.html
DOI: 10.4267/2042/44991

Clinics and pathology

Disease

Rhabdomyosarcomas, the most common pediatric soft tissue sarcomas, are tumours related to the skeletal muscle lineage. The 2 major subtypes are alveolar rhabdomyosarcoma (ARMS) and embryonal rhabdomyosarcoma (ERMS). Other subtypes are botryoid, spindle cell, anaplastic, pleomorphic, and undifferentiated RMS.

Note

Most ARMS cases are characterised by either a t(2;13)(q35;q14), resulting in a PAX3/FOXO1 hybrid gene, or a t(1;13)(p36;q14) resulting in a PAX7/FOXO1 hybrid gene. Most ERMS are characterized by chromosome gains and a loss of heterozygocity in 11p15.

Epidemiology

Three cases of RMS with t(11;22)(q24;q12) have been described to date, including a two-years-old girl with a mixed embryonal and alveolar RMS, who died 14 months after diagnosis, a 4.5-year-old girl, also with a mixed embryonal and alveolar RMS, who was alive and well 9 months after diagnosis (Sorensen et al., 1993; Thorner et al., 1996).

Genetics

Note

A t(2;13) hybrid transcript was excluded in the two cases described by Thorner et al., 1996. In the 4.5-year-old girl case, a highly abnormal karyotype was found, with 85 to 200 chromosomes per mitosis, and MDM2 was amplified more than a hundred times.

Genes involved and proteins

FLI1

Location

11q24

Protein

From N-term to C-term: a 5' ETS domain, a Fli-1-specific transcriptional activation domain, and a 3' ETS transcriptional activation domain. Member of ETS transcription factor gene family. FLI1 binds to DNA in a sequence-specific manner.

EWSR1

Location

22q12

Protein

From N-term to C-term: a transactivation domain (TAD) containing multiple degenerate hexapeptide repeats, 3 arginine/glycine rich domains (RGG regions), a RNA recognition motif, and a RanBP2 type Zinc finger. Role in transcriptional regulation for specific genes and in mRNA splicing.

Result of the chromosomal anomaly

Hybrid Gene

Description

5' EWSR1 - 3' FLI1. EWSR1 exon 7 is fused in frame to FLI1 exon 6.

Fusion Protein

Description

Fusion of the N terminal transactivation domain of EWSR1 to the ETS type DNA binding domain of FLI1.

References

Sorensen PH, Liu XF, Delattre O, Rowland JM, Biggs CA, Thomas G, Triche TJ. Reverse transcriptase PCR amplification of EWS/FLI-1 fusion transcripts as a diagnostic test for peripheral primitive neuroectodermal tumors of childhood. Diagn Mol Pathol. 1993 Sep;2(3):147-57

Thorner P, Squire J, Chilton-MacNeil S, Marrano P, Bayani J, Malkin D, Greenberg M, Lorenzana A, Zielenska M. Is the EWS/FLI-1 fusion transcript specific for Ewing sarcoma and peripheral primitive neuroectodermal tumor? A report of four cases showing this transcript in a wider range of tumor types. Am J Pathol. 1996 Apr;148(4):1125-38

This article should be referenced as such:

Huret JL. t(11;22)(q24;q12) in rhabdomyosarcomas (RMS). Atlas Genet Cytogenet Oncol Haematol. 2011; 15(3):300-301.