

## **Molecular diagnosis of synovial sarcoma: A comparative study with histologic correlation in Ramathibodi Hospital**

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### **ABSTRACT**

**Objective:** Synovial sarcoma (SS) is a relatively common sarcoma of soft tissue arising on extremities of young adult, which is often diagnosed by histology and immunohistochemistry. Synovial sarcoma can have various locations and may cause diagnostic dilemma in tumors arising on unusual locations. In this study, we evaluate the application of molecular test by detection of *SS18/SSX* fusion transcripts as an additional diagnostic tool for synovial sarcoma.

**Method:** We performed the reverse transcription polymerase chain reaction (RT-PCR) to detect the presence of *SS18/SSX* fusion transcripts in 22 tumors from various anatomical sites previously diagnosed as SS or having SS in the differential diagnoses, from year 2010 to 2014.

**Result:** Of the total 22 cases analyzed, 14 cases (63.6%) were positive for *SS18/SSX1* or *SS18/SSX2* fusion transcripts by RT-PCR. The remaining 8 cases (36.4%) were negative for both *SS18/SSX1* and *SS18/SSX2* fusion transcripts. Among 14 cases with positive molecular testing results, 12 cases were previously diagnosed as SS (85.7%), whereas other 2 cases were originally diagnosed as other types of sarcoma (14.3%). On the other hand, from 17 cases which were initially diagnosed as SS by histology and IHC, 12 cases (70.5%) were positive, whereas 5 cases were negative for molecular testing. The diagnoses of *SS18/SSX* fusion negative cases were discussed.

**Conclusion:** We found that the detection of *SS18/SSX* fusion transcripts by RT-PCR is a valuable method to confirm the diagnosis of SS, especially in those difficult cases arising on uncommon sites or unusual histology and unconventional immunohistochemical profiles.

**Keywords:** Synovial sarcoma, *SS18/SSX* fusion transcripts, RT-PCR