Case 398

5. All

【Progress】

His family and he hoped for conservative care, not aggressive treatment of surgical resection and heavy chemotherapy.

【Discussion】

Neurofibromatosis (NF) is currently categorized into three types: NF1, NF2, and Schwannomatosis. NF1 is classically identical to von Recklinghausen disease which is now clinically defined to own more than 6 or more café-aul spots, two or more fibromatoses including iris nodules and/or scoliosis (1-3). NF2 is known to have bilateral acoustic neurinoma, spinal neurinoma/meningioma, cataract and skin pigmentation (4). Schwannomatosis implies multiple schwannomas (5).

Human being owns 23 twin-chromosomes including sex chromosomes. NF1 has abnormality on NF1 gene of Chromosome 17, while NF2 does it on NF2 gene Chromosome 22. NF2 has a single abnormality on Chromosome 22, while Schwannomatosis has 2 or more abnormality on Chromosome 22 (1-4). NF1 and NF2 genes produce cancer suppression proteins of neurofibromin and merlin, respectively. Besides that, Neurofibromin plays the role of producing protein necessary to retain cognition. Abnormality of these genes induce susceptibility of emerge of neurogenic tumors: neurinoma and/or neurofibroma. Neurofibromin shortage induces cognition impairment. These abnormalities inherit as dominant autosomal, inducing 50% incidence of birth with neurofibromatosis (1,6). Neurofibromatosis can occur as a mutation.

NF1 occurs in the incidence of 1/3000, 350000 in Japan, 20/60000 in Hannan city. Some patients become to have bulging skin mass called plexiform of neurofibromatosis (1, 6, 7). Two percent of NF1 is reported to become malignant called malignant nerve sheath tumor in the life-process of NF1: 0.4-person percentage in Hannan city.

Malignant peripheral sheath tumor is characteristic of peripheral parenchyma enhancement on Gd-enhanced MRI or contrast-enhanced CT. Necrosis or cystic formation inside the tumor and peri-tumor edema are also malignancy signs. Two of the three signs (peripheral enhancement, peritumor edema, necrosis or cyst formation inside the tumor) on CT or MRI indicate malignant sign of malignant peripheral sheath tumor (7-13).

Treatment of NF1 for prevention of malignancy is surgical resection at the stage of plexiform of neurofibromatosis. Malignant nerve sheath tumor is apt bleeding during or after surgery. Based on my personal experiences, I have two cases with NF1 and malignant nerve sheath tumor who passed away for postresection bleeding despite of transcatheter arterial embolization. Oral administration of selumetinib is being administrated for the inhibition of progression of NF1 in adolescents of age 3 to 18 (7).

In our patient, he had a huge mass at the left thigh including huge parenchymal solid component, compatible with malignant peripheral sheath tumor.

【Summary】

We presented an eighty-year-old male who was transported to our hospital for fever, dyspnea and motile difficulty. He had café café-aul spots and neurofibroma nodules. A huge mass at right thigh including a solid component, compatible with malignant nerve sheath tumor. It is borne in mind that neurofibromatosis is categorized into three types: NF1(von Recklinghausen disease) NF2 (bilateral acoustic neurinomas) and Schwannomatosis. They are devoid of nerve-cancer-suppression protein, inducing emergence of multiple neurofibroma or neurinoma. Malignant nerve sheath tumor emerges from NF1 whose image characteristics are peripheral parenchyma enhancement on Gd-enhanced MRI or contrast-enhanced CT, including necrosis or cystic formation inside the tumor and peri-tumor edema.

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