神經纖維瘤症第二型

Neurofibromatosis Type 2

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摘要

神經纖維瘤症第二型(neurofibromatosis type 2)又稱爲中樞性神經纖維瘤症,是一罕見的自體顯性遺傳(autosomal dominant)疾病，其致病的機轉可能是位於第22對染色體長臂上(22q12)的腫瘤抑制基因(tumor suppressor gene)產生突變所造成，臨床症狀主要以耳鳴、聽力障礙、暈眩感(vertigo)、顏面神經麻痺、複視等症狀來表現，嚴重的可能會有失明、大腦水腫甚至可能因腫瘤壓迫到腦幹而導致死亡。本院於2002年7月及10月各經歷了1名病患，2人爲母女關係，母親因雙耳耳鳴、聽力障礙，女兒因右側突發性耳聾至本院來求診，2人經純音聽力檢查、言語辨明測驗、聽性腦幹反應檢查，檢查結果懷疑有耳蝸後病變(retrocochlear pathology)，於是安排核磁共振影像檢查，證實2人雙側內聽道均有聽神經瘤，屬於神經纖維瘤症第二型患者，經與病人解釋病情後，2病人均拒絕基因篩檢及手術治療，2病人自行轉院接受左側立體定位珈瑪放射治療，半年後追蹤2病人的聽力均有受損，核磁共振影像檢查顯示2病人腫瘤大小未因珈瑪放射治療而變小。由於神經纖維瘤症第二型爲罕見的病例，其發生機率爲四萬分之一，特此提出報告，並就過去的文獻，討論此病的成因、診斷及處置等。

Abstract

Neurofibromatosis type 2 (NF-2), also called central type neurofibromatosis, is a rare autosomal dominant disease caused by a mutated tumor suppressor gene on the long arm of chromosome 22q12. The prominent clinical symptoms include tinnitus, hearing impairment, vertigo, facial palsy, diplopia, etc. The severe results can be blindness, hydrocephalus, or even fatal tumor-induced brainstem compress ion. In July and October 2002, respectively, a mother visited our department for tinnitus and hearing impairment, and so did the daughter for right sudden deafness. Thus the pure tone audiometry, speech discrimination test, auditory brainstem reflex examination were executed for them both. The results revealed the retrocochlear pathology, and magnetic resonance imaging was arranged. Attributed to the bilateral vestibular schwannomas detected, NF-2 was diagnosed. After the disease was explained, the screening of genes and surgical intervention were refused, and they went to another hospital where gamma knife stereotactic radiosurgery for their left s ides was implemented. After half year by our follow up, their hearing impairment was noted, and magnetic resonance imaging indicated the same tumor size which was not reduced by the radiosurgery. Due to the rarity of the disease, the incidence rate was

approximately 1/40000. The case with the causes, diagnoses and treatments discussed all according to the literature review is hereby presented