Genetic Counselling and Education

# AB065. Genetic testing and counseling in family with late onset autosomal dominant spinocerebellar ataxia

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**Background:** Spinocerebellar ataxia (SCA) is neurodegenerative disorders with autosomal dominant inheritance, characterized by progressive ataxia. More than 30 types of SCA are known caused by various causative genes. SCA3 (*MJD1* gene) is the most common form of SCA. We present an SCA3 family with complicated genetic counseling issue.

**Methods:** A 44-year-old female was referred to our service for genetic consultation. Pedigree construction, physical examination, and gene mutation analyses were performed on specimens from the patient and some family members. CAG repeat analysis of ataxin1 (SCA1), ataxin2 (SCA2), *MJD1* (SCA3), and *CACNA1A* (SCA6) genes were done, followed by fragment length analysis, and sequencing. Genetic counseling was provided.

**Results:** The patient was already bedridden and she had a brother with the same condition. Neurological examination showed multiple cranial nerve palsy, right eye twitching, spastic paraplegia, limb atrophy, numbness, and bowel-bladder incontinence. CAG repeat expansion (>44) of *MJD1* gene was found (28/76 repeats alleles), confirming SCA3. As requested by her family, carrier testing was done for her 15-year-old daughter, 12-year-old nephew, and 10-year-old niece. Her daughter had CAG repeat expansion (27/77 repeats alleles), while the nephew and niece revealed normal alleles. Problem arose when patient died prior to mutation analysis was complete and her husband divorced, leaving the daughter being an orphan. Risk and consequences of positive testing were explained to her uncle, who decided to keep information until the child would have reached legally adult age.

**Conclusions:** Genetic counseling is needed, especially in situation which involves many affected members. Carrier testing ethically should be taken for adult who can signs consent for themselves and should be discouraged for under aged individuals. Nevertheless, test was done as parent's request, because of family anxiety and curiosity. Complexity and adult onset of SCA remain as challenges in providing carrier testing and genetic counseling.

**Keywords:** Genetic counseling; spinocerebellar ataxia (SCA); carrier testing

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