**Marinoresco-Sjögren syndrome (MSS)**

- Congenital cataracts
- Ataxia with cerebellar atrophy
- Mental retardation
- Myopathy
- Causative gene: SIL1 on Ch.5q31
- SIL1 involves in protein quality control

**METHODS**

★ A nationwide survey of MSS patients

Questionnaires were sent to 5,452 Japanese neurologists and child neurologists. Detailed clinical information and follow-up data of individuals with possible MSS were obtained.

★ Mutation screening

Direct sequencing of SIL1 was performed using genomic DNA.

★ Zebrafish model of MSS

SIL1 knockdown zebrafish was produced by morpholinos (MO). Skeletal muscle, eyes, and cerebellum were examined.

**RESULTS**

- A homozygous c.937dupG was identified in 21 of 24 (87.5%) individuals with SIL1 mutation.
- Three patients with no mutation in SIL1 had similar clinical features.

**CONCLUSIONS**

- The life prognosis of MSS appears to be good.
- Respiratory, cardiac, and swallowing functions are well preserved, even in the patients who are over 50 years of age.
- Zebrafish is a good animal model to screen for potential therapeutic drug candidates.

**REFERENCES**