■ Clinical commentary

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Epileptic Disorders

Possible critical region associated with late-onset spasms in 17p13.1–p13.2 microdeletion syndrome: a report of two new cases and review of the literature

Naohiro Yamamoto[1], Shin Okazaki[1], Ichiro Kuki[1], Naoki Yamada[1], Shizuka Nagase[1], Megumi Nukui[1], Takeshi Inoue[1], Rie Kawakita[2], Tohru Yorifuji[2], Takao Hoshina[3] Toshiyuki Seto[3], Toshiyuki Yamamoto[4,5], Hisashi Kawawaki[1]

[1] Division of Pediatric Neurology, Osaka City General Hospital, Osaka, Japan
[2] Division of Pediatric Endocrinology and Metabolism, Osaka City General Hospital, Osaka, Japan
[3] Department of Pediatrics, Osaka City University Graduate School of Medicine, Osaka, Japan
[4] Tokyo Women's Medical University Institute for Integrated Medical Sciences, Tokyo, Japan
[5] Institute of Medical Genetics, Tokyo Women's Medical University, Tokyo, Japan

Epileptic Disorders

17p13.1-13.2 microdeletion syndrome

- congenital anomaly syndrome with characteristic facial features and multiple malformations
- is seldom a cause of epilepsy

Late-onset spasms with 17p13.1-13.2 microdeletion syndrome

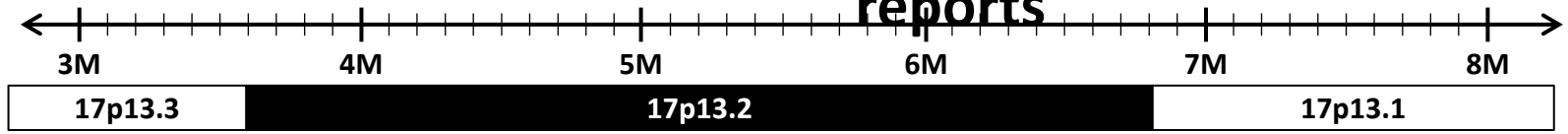
- Only one case reported with late-onset spasms

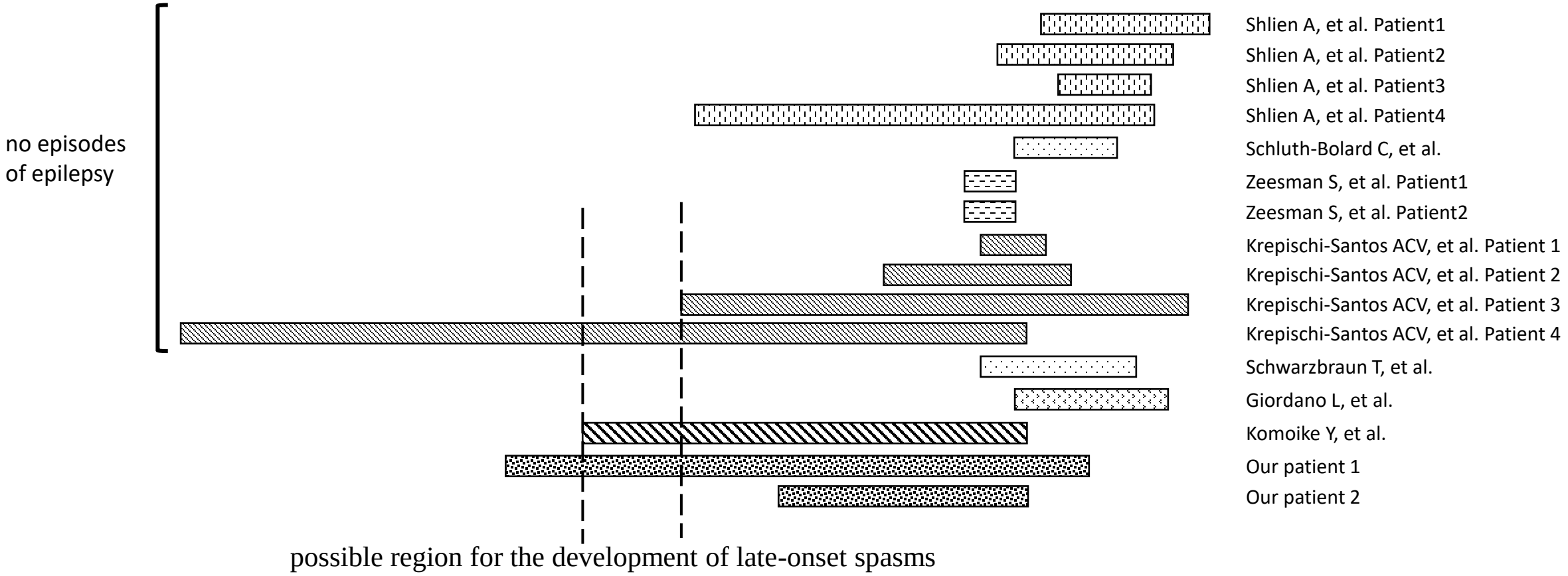
In this case report we discuss

- the possible region in the development of late-onset spasms

Epileptic Disorders

The locations of the deleted regions and epilepsy-related genes in previous reports

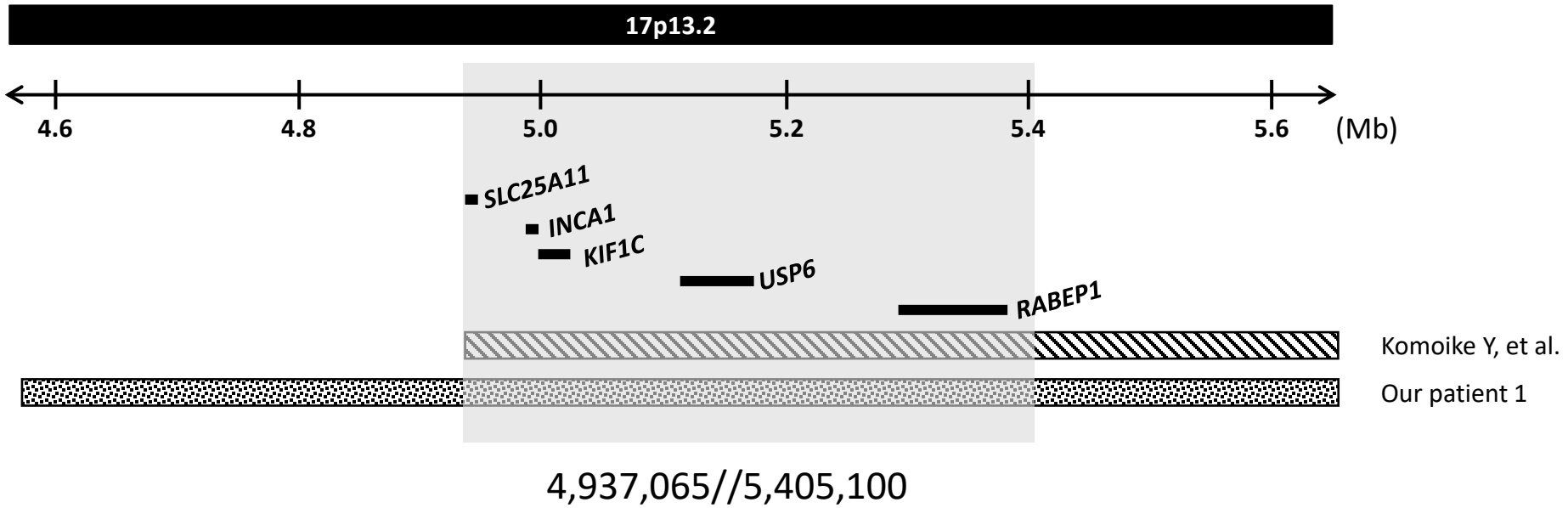




- We considered the possibility that the common mutation region, excluding the mutation site of the case with 17p13.1-13.2 microdeletion in which no epilepsy was reported, might be involved in the development of late-onset spasms.

Epileptic Disorders

Possible region involved in the development of late-onset spasms



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- A decrease in NMDA receptor expression, NMDA receptor destabilization or accumulation of neurotoxic substances caused by the *USP6* gene deletion might be involved in the development of late-onset spasms