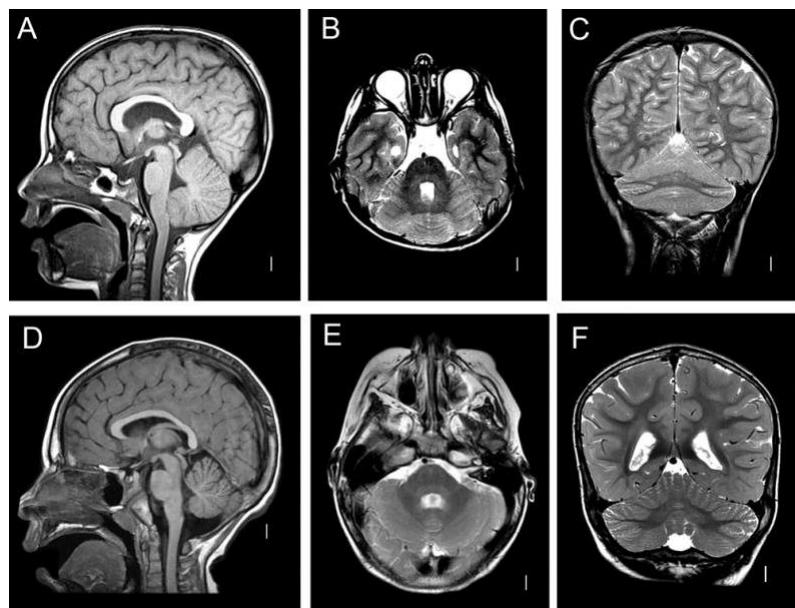


Tully HM, et al. Persistent figure-eight and side-to-side head shaking is a marker for rhombencephalosynapsis. Mov Disord 2013;28:2019-23.



1

Video 1: 21 month old boy with head shaking.



2

1

Video 2: 3 year old boy with head shaking. Note the cessation of movements with visual fixation.



くつ下はきと関連



右上を見る
→正面からの入力は上方網膜に入る



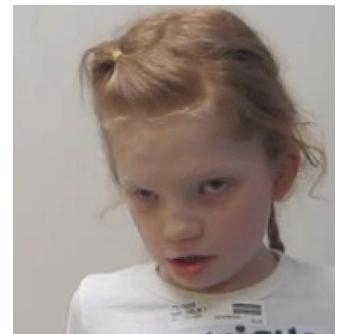
少し頸後傾で上を見る
→正面からの入力は上方網膜に入る

3

Video 3: 8 year old girl with head shaking.



- 対話と関連
- 上目使いで相手を見る
→非両眼視の上方網膜に入力



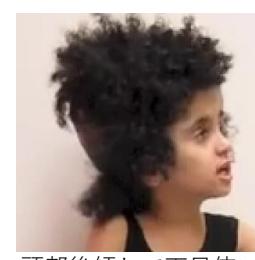
4

2



Video 4: 10 year old girl with head shaking. Note additional stereotypies (hand flapping) and cessation of movements with visual fixation.

➤ 左右にいる人と対面と関連



- 閉眼した後に見る

• 頭部後傾して下目使いで見る
→非両眼視の上方網膜に入力



- 頸部右傾左回旋位で右方を見る
→右眼鼻側網膜上方に入力

5



Video 5: 27 year old woman with head shaking. This patient had been given a diagnosis of aqueductal stenosis in infancy. A clinician recognized the relationship between headshaking and hindbrain malformations, reviewed her MRI, and discovered previously unrecognized rhombencephalosynapsis.

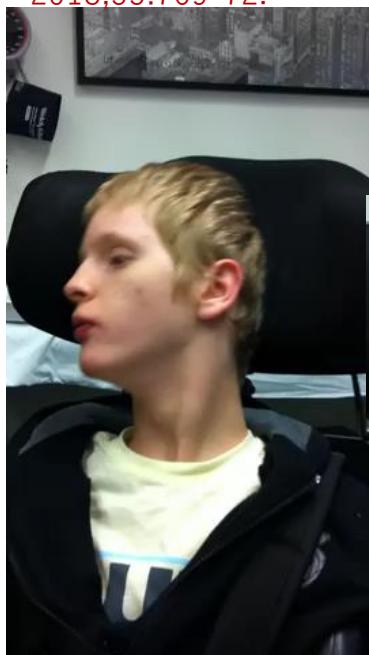


- 頭を右下から上に振り上げる
下向き薄目で見ている？

6

3

Kim YO, et al. Head stereotypies in STXBP1 encephalopathy. Dev Med Child Neurol 2013;55:769-72.



Video S1. Continuous, 1Hz, rhythmic, figure-of-eight head stereotypy and dyskinetic hand movements that briefly settled during a period of concentration (patient 1 at 13 years).

➤ 頭部後屈と回旋の共存



- 頭後傾で左斜め下を見ている
→正面からの入力は左眼鼻側網膜上方で受ける



Video S2. Truncal stereotypies rocking constantly from side-to-side in concert with the slow figure-of-eight head and hand stereotypies when lying on the floor (patient 1 at 14 years).

7

Video S3. Figure-of-eight head stereotypy and dyskinetic hand movements that increased with excitement but settled with concentration and was associated with smiling and sometimes vocalization (patient 2 at 9 years).



- 右方を頸後屈で正面視し、下目使いで見る
→非両眼視の上方網膜に入力

8

4

Video S4. A fast (2–3Hz) head stereotypy of a side-to-side 'no' movement occurring in isolation or in conjunction with complex right upper limb stereotypy where she flicked her ear with her right hand (patient 3 at 11 years).



速い首振り→正面遠方のものは不動であり、その前方のものは大きく動く
↑見えてくる

9



Stamberger H, et al.
Natural History Study of
STXBP1-Developmental
and Epileptic
Encephalopathy Into
Adulthood. Neurology
2022;99:e221-e233.

Video showing 4 adult
patients with STXBP1-
DEE with mild to
pronounced gait
abnormalities

- (1): a 21-year-old man with mildly broad-based gait with external rotation of the feet and pes planus;
- (2): a 19-year-old woman with more pronounced broad-based gait with external rotation and eversion of the feet while walking and pes planus;
- (3): a 20-year-old man with pronounced ataxic gait with external rotation of the feet;
- (4): a 22-year-old woman with broad-based gait with asymmetric posturing and dystonia of the left arm and impression of hypertonia in the legs.

10

Case 1: 20-year old woman showing hand stereotypies and figure-of-eight head stereotypies



- 頭後傾で右斜め下を見ている
→正面からの入力は右眼
鼻側網膜上方で受ける



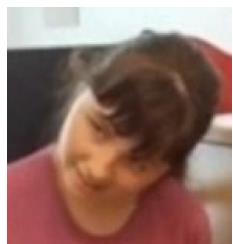
右上から左下へ頭を振り下げる

Video 4

Stereotypies including figure-of-eight head stereotypies in 3 adult patients with STXBP1-DEE.

11

Case 2: 18-year old woman showing hand and figure-of-eight head stereotypies and body rocking



右上方を見る



左側方を見る

✓ どこを見ているかわからないが、
正面視をしないことは確かである

Video 4

Stereotypies including figure-of-eight head stereotypies in 3 adult patients with STXBP1-DEE.

12

Case 3: 19-year old man showing figure-of-eight head stereotypies



✓下目使いの8の字運動もあり

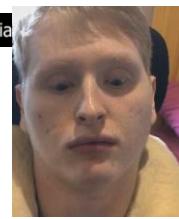


Video 4
Stereotypies including figure-of-eight head stereotypies in 3 adult patients with STXBP1-DEE.

Case 1: 19-year old man with hand and face dyskinesia



Video 5
Dyskinesia including choreiform movements in 2 patients with STXBP1-DEE.



頸後屈で眼瞼下降しない
眼球下転（落陽現象）

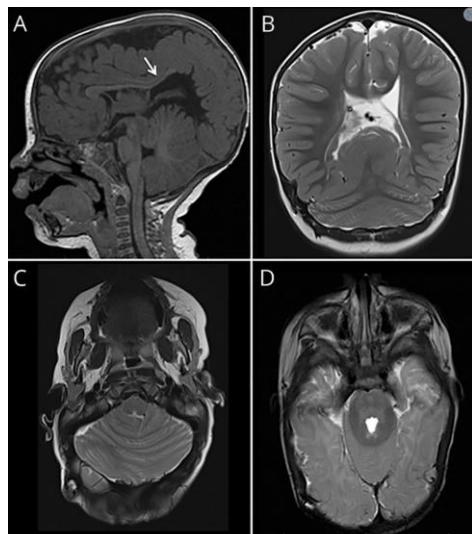
- 正面から上方網膜に入力



横地の眼球下転との違いは
・上眼瞼挙筋の常時筋収縮状態が、下方視に伴う眼輪筋の収縮に打ち勝つ

13

Accogli A, Srour M. Teaching Video Neurolimages: Figure 8 head-shaking stereotypy in rhombencephalosynapsis. Neurology 2018;90:e1832-e1833.



14

Poretti A, et al. Horizontal head titubation in infants with Joubert syndrome: a new finding. Dev Med Child Neurol 2014;56:1016-20.



側方視をしている

Video S1: A 2-month-old female with Joubert syndrome and head titubation (patient 1). Note the high frequency and small amplitude of the head titubation.



➤ Joubert症候群の幼児期にみられる速い短時間の首振りの原型

運動視差による奥行き知覚
・近くのものは大きく動く
・遠くのものは小さく動く

Video S2: A 2-month-old male with Joubert syndrome and head titubation (patient 5).

15



Video S3: A 12-month-old male with Joubert syndrome and head titubation (patient 12).

16

Sakurai Y et al. Head titubation and irritability as early symptoms of Joubert syndrome with a homozygous NPHP1 variant. Brain Dev 2021;43:863-866.



17

Delorme C et al. Spasmus Nutans: More Than Meets the Eye. Pediatr Neurol 2015;53:367-8.

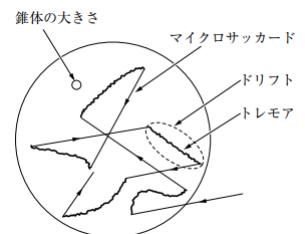


- 側方視
- 首振り
- 眼振

Fixational eye movement

- microsaccade
- drift
- tremor

は正常に発達してはいないだろう
このovershootが眼振か



18

Sala-Coromina J, et al. Early-onset eyelid stereotypies are a frequent and distinctive feature in Dravet syndrome. Seizure 2021;92:155-157.

Patient 1
32 months old

- 目つぶり → 視覚リセット
- 頸後屈し、左方視が主体



Patient 1
10 years old

- 閉眼・目つぶり～薄目・下方視
- 首振り



19

Patient 2
6 years old

薄目・下目使い or 発作？



Patient 3
18 months old

目つぶり → 視覚リセット



20

10

Patient 4
13 months old

目つぶり → 視覚リセット
正面視している



21

まとめ

- 発達期脳障害で、常的にみえる大半の頸運動は、視覚行動である
- たいてい中心視はせず、周辺視をしている
- それで、運動視差とoptic flowによる奥行き知覚を得ている
- 目つぶりは視覚リセットである