P2-189 Anophthalmia, micrognathia, combined pituitary hormone deficiency, severe growth retardation and liver dysfunction induced levothyroxine sodium powder in a boy with microdeletion of 14q22q23

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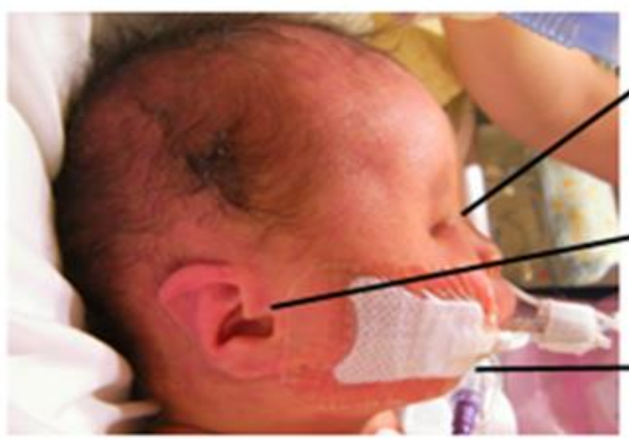
CONFLICT OF INTEREST
<Satomi Koyama>
I declare that I have no potential conflict of interest.

Introduction

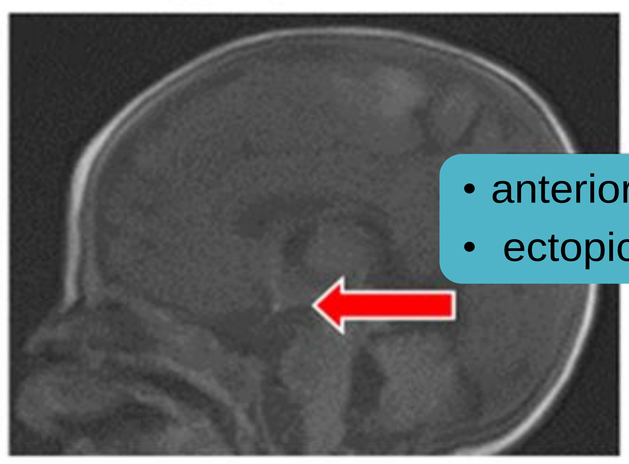
- Microdeletion of 14q22q23 results in a rare chromosomal disorder associated with microphthalmia/anophthalmia, pituitary anomalies, polydactyly/syndactyly, micrognathia, growth restriction and mental retardation.
- Haploinsufficiency of the genes OTX2 (orthodenticle homeobox 2) and BMP4 (bone morphogenetic protein 4) are responsible for most of the phenotypic features in the 14q22q23 microdeletion syndrome.
- There are only a few reports about liver dysfunction induced by levothyroxine in childhood.

Case

- The patient was born at 38 weeks and 5 days. His birth length and weight were 41.5cm (-3.5SD) and 1946g (-2.5SD). He showed bilateral anophthalmia, micrognathia, low set ears, micropenis, cryptorchidism at birth. He also had respiratory failure, hypoglycemia and bilateral hearing loss.



The brain magnetic resonance imaging

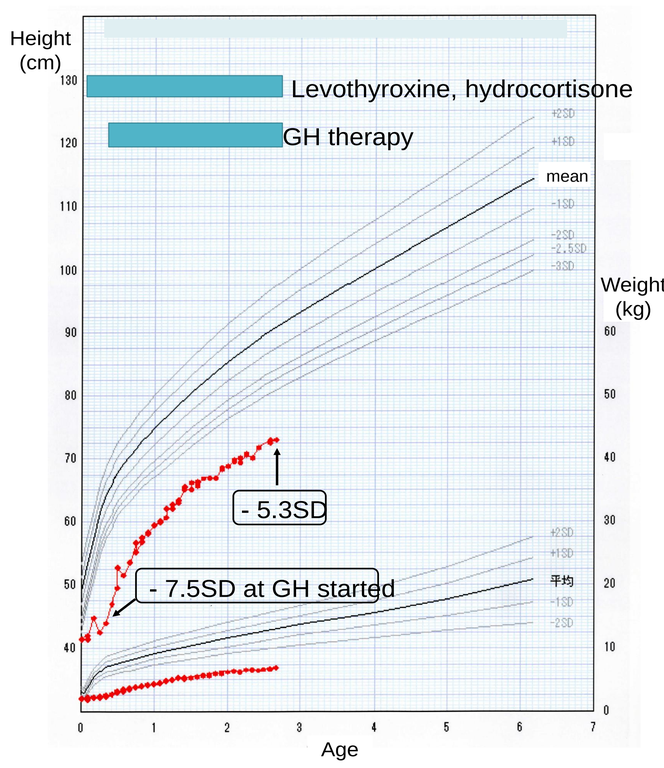


【Endocrine examination (day 26)】

FT3 2.96 pg/ml	ACTH 16.1 pg/ml
FT4 0.74 ng/dl (↓)	Cortisol 1.0 μg/dl (↓)
TSH 1.92 μIU/ml	LH ≦0.07 mIU/ml(↓)
IGF-1 11 ng/ml (↓)	FSH ≦0.30 mIU/ml (↓)
Arginine stimulation test (day82)	Testosterone ≦0.03 ng/ml (↓)
GH 1.3→1.2(60min) ng/ml (↓)	PRL 7.80 ng/ml

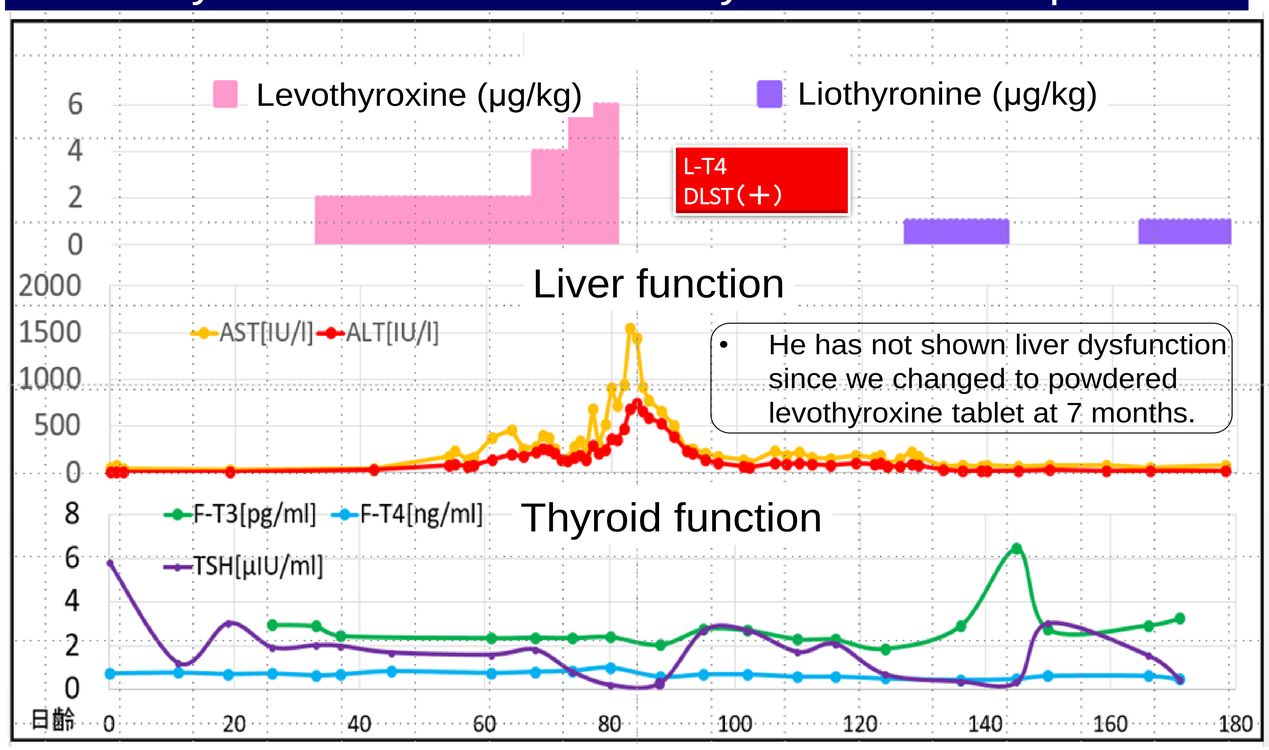
【Karyotype】 46,XY
【array CGH】 7.6Mb microdeletion of 14q22.1q23.1

Growth chart

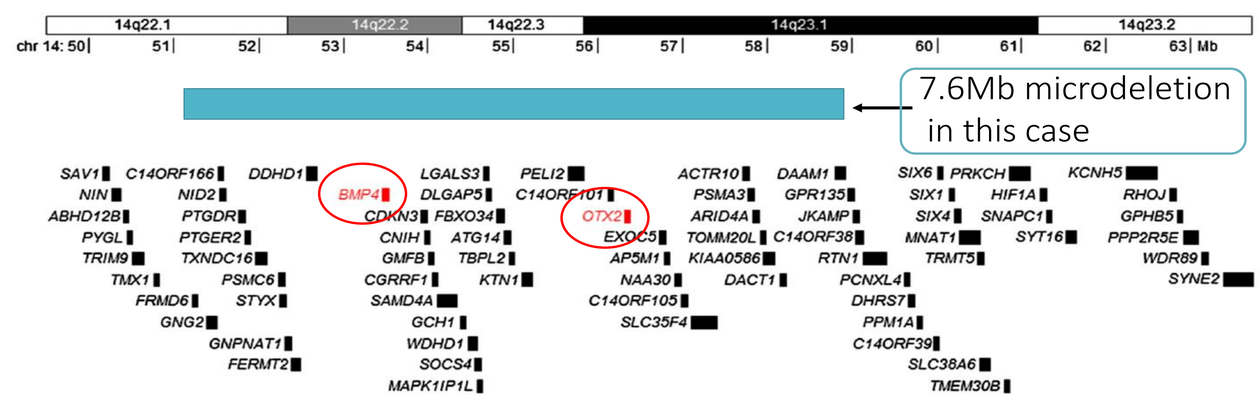


- Levothyroxine sodium powder and hydrocortisone administration started at 1 month and GH therapy started at 4 months.
- His height SD was -7.5 at 4 months and -5.3 at 2 years and 7 months.

Liver dysfunction induced levothyroxine sodium powder



Microdeletion of 14q22q23



Conclusion

- GH therapy slightly improved growth rate in this case, but since GH deficiency was probably not the only factor responsible for growth retardation, in patients with 14q22q23 deletion, GH therapy was not completely effective in stimulating normal growth.
- This finding is not unexpected because previous reports showed there were patients with growth retardation in the absence of GH deficiency and growth was difficult to correct with GH therapy in patients with GH deficiency[1,2].

【References】
1) Brisset S et al. Molecular Cytogenetics 2014, 7:17
2) Nolen LD et al. Am J Med Genet 2006, 140:1711-1718