Genotype and Clinical Characteristics in Korean patients with Prader-Willi Syndrome

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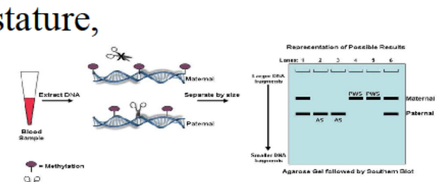
Disclosure statement

The authors have nothing to disclose or conflicts of interests in this study.

Introduction

- Prader–Willi syndrome (PWS) is characterized by neonatal hypotonia, hypogonadism, progressive obesity, short stature, and mental retardation.
- This syndrome arises from a loss of expression of paternally derived genes on chromosome 15q11–13 region.
- The aim of this study was to investigate clinical characteristics and their genotypes in Korean patients with PWS.



Methods & Patients

- The study included 46 PWS patients diagnosed by clinical features and methylation test. And genetic subtypes were distinguished by using fluorescence in situ hybridization (FISH).
- Birth history, auxological profile, clinical features, neurodevelopmental state, radiologic findings, and medication were reviewed, retrospectively. And body mass index (BMI) was analyzed in patients more than two years old age.

Results

- Total 46 PWS patients
  - ✓ M:F=27:19 patients
  - ✓21 males (77.8%) : s/p orchiopexy
- Age at diagnosis : 1.8 ± 3.7 years.
  - ✓<1yr: 39 patients (84.8%) (hypotonia, feeding difficulty)
  - ✓3~15yr : 7 patients (obesity, developmental delay)
- FISH was performed in 39 patients
  - ✓ 29 paternal deletions (73.7%) ,10 maternal UPDs (26.3%)

Table 1. Genotype and Clinical characteristics of patients with Prader-Willi syndrome (PWS)

Parameter	Total 46 PWS patients (mean ± SD)	Subtype (39 patients)	
		Paternal deletion (29 patients)	Maternal UPD (10 patients)
Male (n, %)	27 (58.7%)	18 (62%)	6 (60%)
Maternal age at birth (years)	31.8 ± 4.4 (range, 24-42)	31.5 ± 4.3 (range, 24-42)	33.2 ± 5 (26-41)
Age at diagnosis (years)	1.8 ± 3.7 (10 days - 15y)	1.3 ± 2.8 (10 days -10y)	2.7±4.9 (15 days - 15y)
Current age (years)	6.82 ± 6.44	6.05 ± 5.06	5.44 ± 4.99
Growth hormone therapy	36 (78.3%)	28 (96.5%)	8 (80%)
Age at initiation of GH therapy (years)	2.7 ± 2.9 (5m - 10y)	2.5 ±2.9 (5m - 10y)	3.1±3.0 (6m - 7.8y)
Duration of GH therapy (years)	3.3 ± 2.4	3.6 ± 2.5	2.2 ±1.7
Height (cm)	110.8 ± 27.4	108.6 ± 28.4	104.8 ± 22.2
Weight (kg)	30.1 ± 21.8	27.3 ± 25.2	25.2 ± 16.5
BMI (kg/m²)	2.3 ± 0.9	0.8 ± 1.5	1.2 ± 2.0
Obesity (n, %)	19 (41.3%)	9 (31%)	6 (60%)
Scoliosis (n, %)	20 (43.5%)	12 (41%)	6 (40%)
Orchiopexy (n, %)	21 (78.3%)	14 (77.8%)	4 (66.7%)
Epilepsy with AED (n, %)	6	6	0
Precocious puberty (n)	3	3	0
Spleep apnea (n)	2	1	1
Congenital hip dysplasia (n)	2	0	2

PWS, Prader-Willi syndrome; SD, standard deviation; UPD, uniparental disomy; GH, growth hormone; BMI, body mass index; AED ,antiepileptic drug.

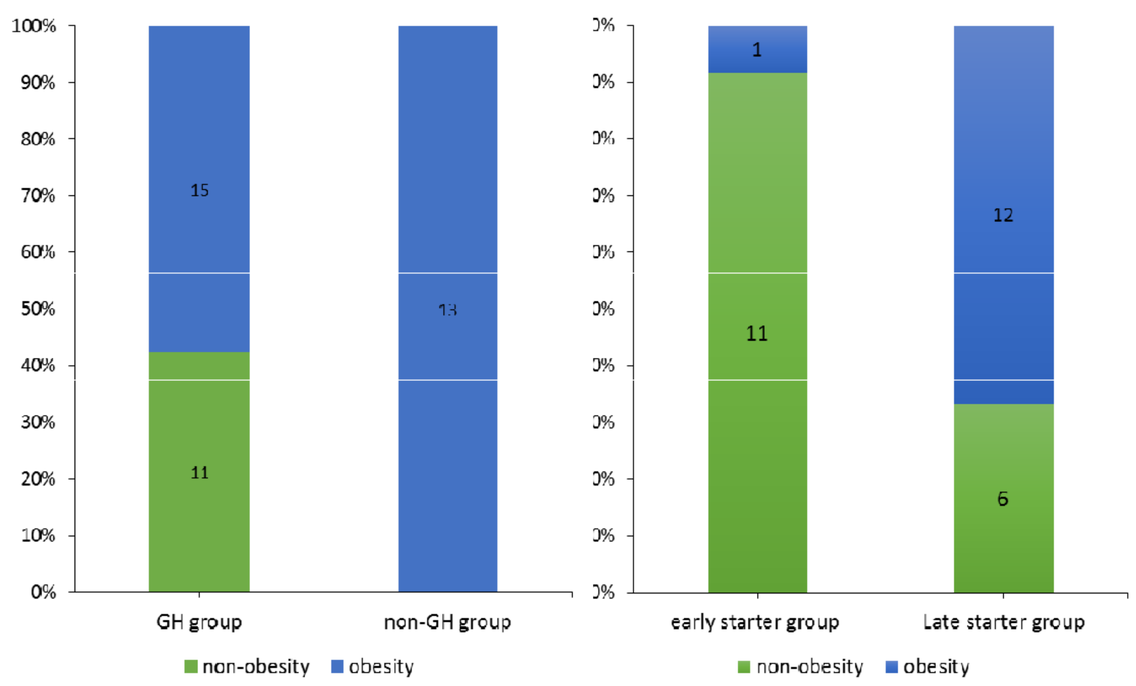


Fig. 1. Comparison of obesity between growth hormone group and non-growth hormone group

- GH hormone therapy
  - ✓36 patients (78.3%), age at the initiation of GH: 2.7 ± 2.9 yr
  - ✓Duration of therapy: 3.3 ± 2.4 years
- Obesity : 19 patients (19/37, 51.4%, age> 2yrs)
- BMI= 2.34 ± 0.86 kg/m²
- GH group was less obese than non-GH group (41.9%, 100%, p <0.05)
- In GH group, there were different proportions of obesity according to timing of GH therapy.
  - ✓Early starter group who had received GH within the age of one had a one obese patient (1/12, 7.7%)
  - ✓Late starter group were more obese (12/18, 66.7%, p <0.05)
- Other characteristics:
  - ✓ Scoliosis : 20 patients (43.5%)
  - ✓ Two patients had hip dysplasia, and one patient had pes planus → 3 patients: matUPD
  - ✓ Epilepsy : 6 patients received anti-epileptic drugs for epilepsy, and all of them were paternal deletion.
  - ✓ Central precocious puberty: 3 patients with paternal deletion
  - ✓ Two patients performed tonsillectomy due to sleep apnea.

Conclusions

- Our study showed that deletion was more common, and early GH therapy improved BMI of Korean PWS patients.
- As PWS is a multi-systemic disorder, and there are different manifestations according to time, persistent and systemic monitoring should be needed.

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Fat 1

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