

Physical assessment in Chinese children with 5 α -reductase type 2 deficiency

Xiu Zhao^{1,9#}, Shaoke Chen^{2#}, Yanning Song¹, Xiumin Wang³, Feihong Luo⁴, Yu Yang⁵, Linqi Chen⁶, Ruimin Chen⁷, Hui Chen⁸, Zhe Su⁹, Di Wu¹, Chunxiu Gong^{1*}

¹ Beijing Children's Hospital, Capital Medical University, Beijing, China

³ Shanghai Children's Medical Center, Shanghai Jiaotong University, Shanghai, China

⁵ Jiangxi Provincial Children's Hospital, Nanchang, China

⁷ Fuzhou Children's Hospital, Fuzhou, China

⁹ Shenzhen Children's Hospital, Shenzhen, China

² Maternal and Child Health Hospital of Guangxi Zhuang Autonomous Region, Nanning, China

⁴ Children's Hospital of Fudan University, Fudan University, Shanghai, China

⁶ Children's Hospital of Soochow University, Suzhou, China

⁸ Capital Medicine University, Beijing, China

Objectives:

To study the growth pattern in Chinese pediatric patients with 5 α RD.

Methods:

Data were from 187 patients with 5 α RD (age from 0-16 years old) who visited 8 pediatric endocrine centers from Jan, 2010 to Dec, 2017. Children with 46, XY DSD without hormone treatment and those with testicular dysfunction were also selected as a positive control group. Data were from 187 patients with 5 α RD (age from 0-16 years old) who visited 8 pediatric endocrine centers from Jan, 2010 to Dec, 2017. Children with 46, XY DSD without hormone treatment and those with testicular dysfunction were also selected as a positive control group.

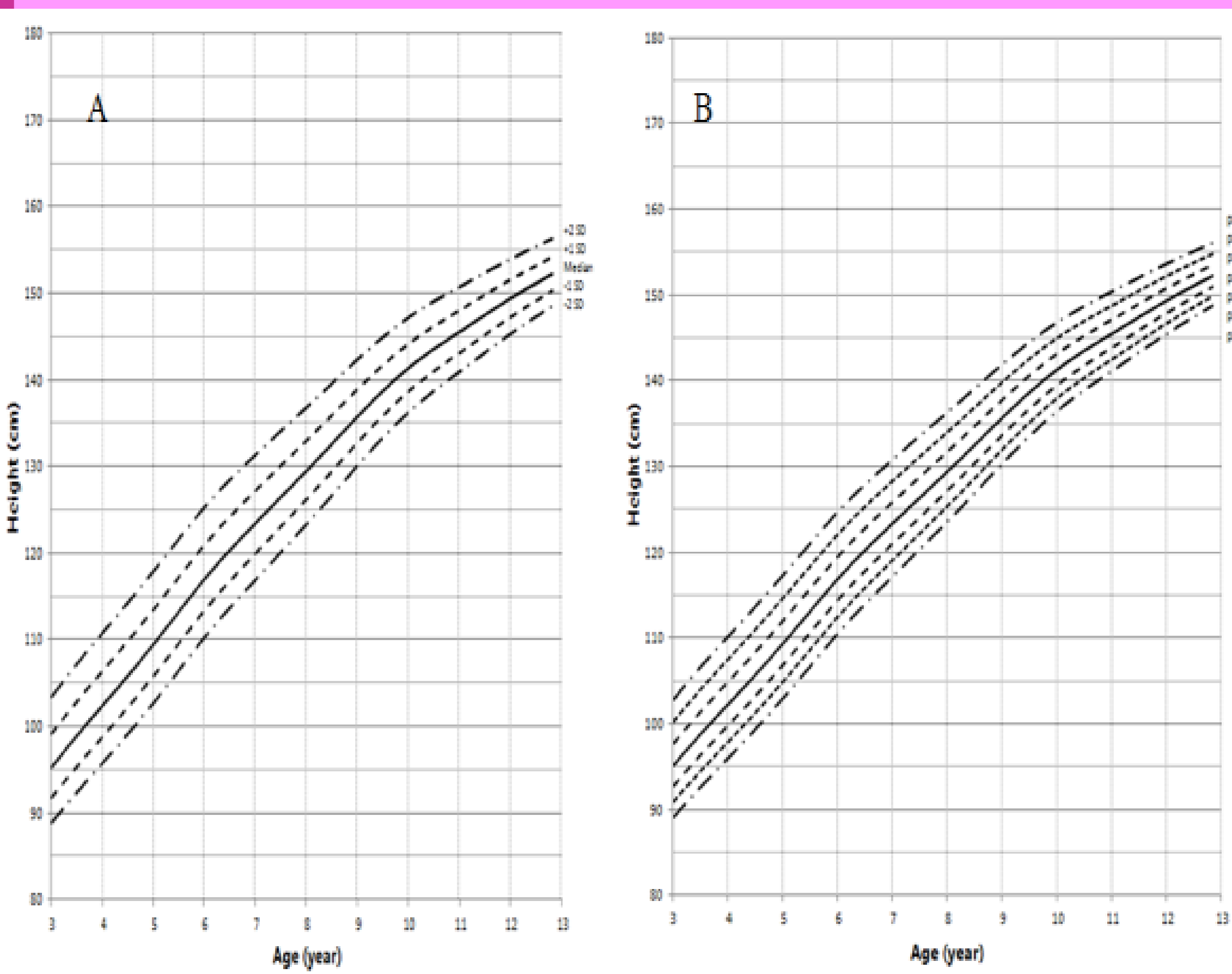


Fig 3. Height in children with 5 α RD (3-12 years old). (A) Percentile curve; (B) Standard deviation curve.



Fig 2. Geographical distribution of children with 5 α RD2 in China.

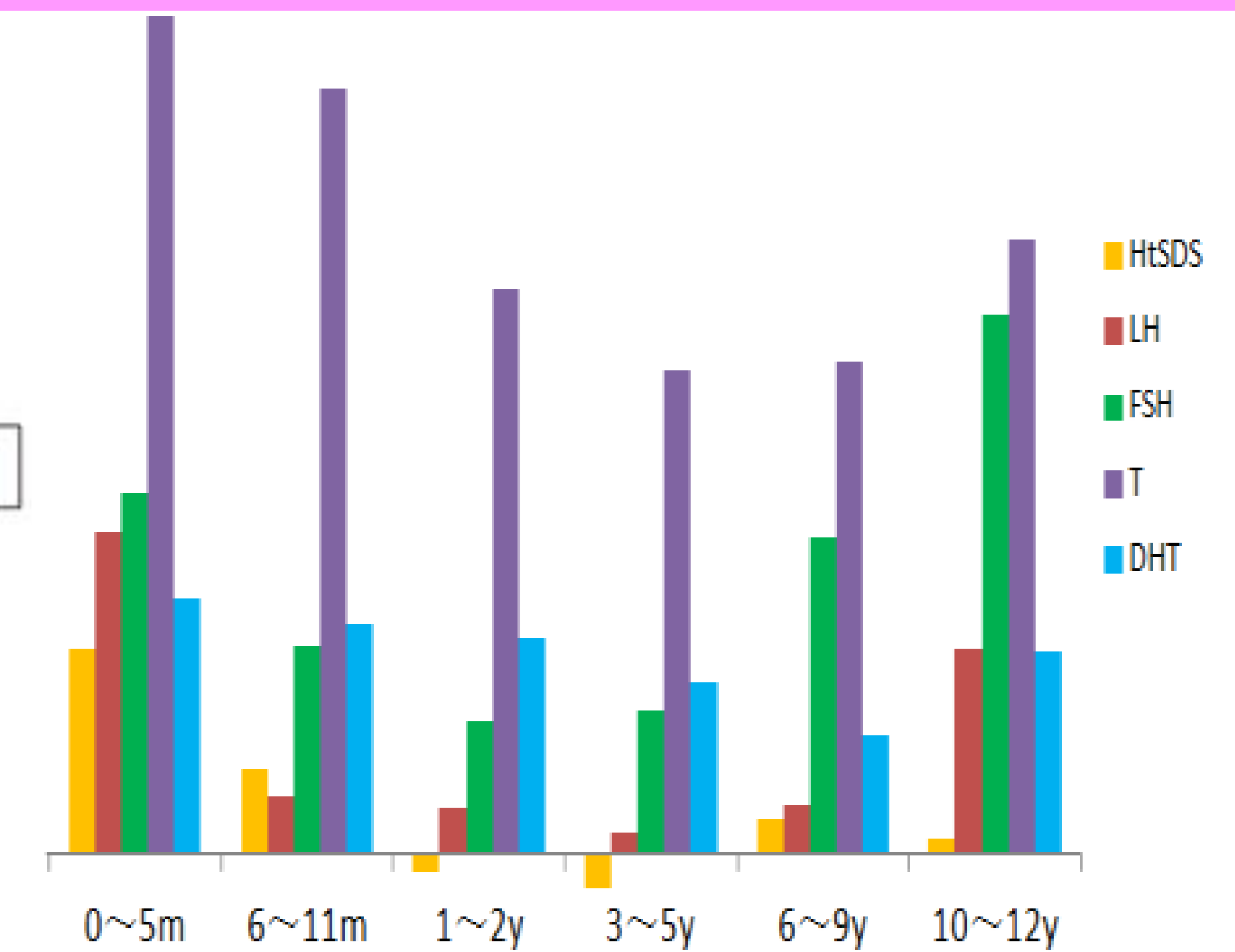


Fig 3. Hormone and HtSDS for Chinese children with 5 α RD among different age groups. Yellow is HtSDS (height standard deviation score). Red is LH (luteinizing hormone). Green is FSH (follicle-stimulating hormone). Purple is T (testosterone). Blue is DHT (dihydrotestosterone). T and DHT were data after HCG stimulation test.

Results:

Compared to normal boys, higher height standard deviation scores (HtSDS) were observed in 5 α RD children who were <1-year-old ($p=0.002$, 0.048 , respectively), and higher weight standard deviation scores (WtSDS) in those <6-month-old ($p=0.012$). Then HtSDS and WtSDS showed lower than those of normal boys of the same age when >2-year-old. The variation tendency of HtSDS in Chinese 5 α RDs was consistent with the trend of T after HCG stimulation test. HtSDS and T after HCG stimulation test in the EMS <7 group were higher than those in the EMS ≥ 7 group. Additionally, the ratio of bone age over chronological age (BA/CA) was lower than 1 in 5 α RD children.

Conclusions:

The children with 5 α RD had a special growth pattern which was affected by high level of T. Their body length was longer in 0~5 months group, and then its growing slowed down leaving children shorter than normal boys after the age of 2 years old. The bone age was delayed in 5 α RD children. This may provide a chance for androgens treatment in young age 5 α RD boys for their micropenis.

References:

1. Isojima T, Yokoya S, Ito J, Horikawa R, Tanaka T 2009 New reference growth charts for Japanese girls with Turner syndrome. PEDIATR INT 51:709-714
2. Angulo MA, Castro-Magana M, Lamerson M, Arguello R, Accacha S, Khan A 2007 Final adult height in children with Prader-Willi syndrome with and without human growth hormone treatment. AM J MED GENET A 143A:1456-1461
3. Wollmann HA, Schultz U, Grauer ML, Ranke MB 1998 Reference values for height and weight in Prader-Willi syndrome based on 315 patients. EUR J PEDIATR 157:634-642
4. Lyon AJ, Preece MA, Grant DB 1985 Growth curve for girls with Turner syndrome. ARCH DIS CHILD 60:932-935
5. Hughes IA, Northstone K, Golding J 2002 Reduced birth weight in boys with hypospadias: an index of androgen dysfunction? Arch Dis Child Fetal Neonatal Ed 87:F150-F151
6. Richter-Unruh A, Knauer-Fischer S, Kaspers S, Albrecht B, Gillessen-Kaesbach G, Hauffa BP 2004 Short stature in children with an apparently normal male phenotype can be caused by 45,X/46,XY mosaicism and is susceptible to growth hormone treatment. EUR J PEDIATR 163:251-256
7. Han TS, Goswami D, Trikudanathan S, Creighton SM, Conway GS 2008 Comparison of bone mineral density and body proportions between women with complete androgen insensitivity syndrome and women with gonadal dysgenesis. EUR J ENDOCRINOL 159:179-185
8. Di Wu, Gong CX, Qin M 2013 Analysis of the clinical characteristics and body height in 153 disorders of sex development children without known cause. Chinese Journal of Evidence-Based Pediatrics 8:46-49
9. Wu D CHG 2017 Physical assessment and reference growth curves for children with 46,XY disorders of sex development. pediatr Invest 1:16-22
10. Veiga-Junior NN, Medaets PA, Petrolis RJ, Calais FL, de Mello MP, Castro CC, Guaragna-Filho G, Sewaybricker LE, Marques-de-Faria AP, Maciel-Guerra AT, Guerra-Junior G 2012 Clinical and Laboratory Features That May Differentiate 46,XY DSD due to Partial Androgen Insensitivity and 5 α -Reductase Type 2 Deficiency. INT J ENDOCRINOL 2012:964876
11. Ittiwut C, Pratuangdejkul J, Supornsilchai V, Muensri S, Hiranras Y, Sahakitrungruang T, Watcharasindhu S, Suphapeetiporn K, Shotelersuk V 2017 Novel mutations of the SRD5A2 and AR genes in Thai patients with 46, XY disorders of sex development. J Pediatr Endocrinol Metab 30:19-26
12. Sultan C, Paris F, Terouanne B, Balaguer P, Georget V 2001 Disorders linked to insufficient androgen action in male children. HUM REPROD UPDATE 7:314-322
13. Choi JH, Kim GH, Seo EJ, Kim KS, Kim SH, Yoo HW 2008 Molecular analysis of the AR and SRD5A2 genes in patients with 46,XY disorders of sex development. J Pediatr Endocrinol Metab 21:545-553
14. Deeb A, Al SH, Ibukunoluwa F, Attia S 2016 Phenotype, Sex of Rearing, Gender Re-Assignment, and Response to Medical Treatment in Extended Family Members with a Novel Mutation in the SRD5A2 Gene. J Clin Res Pediatr Endocrinol 8:236-240