

The influence of Growth Hormone treatment on Fat-free Mass in Kabuki Syndrome children.

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Introduction

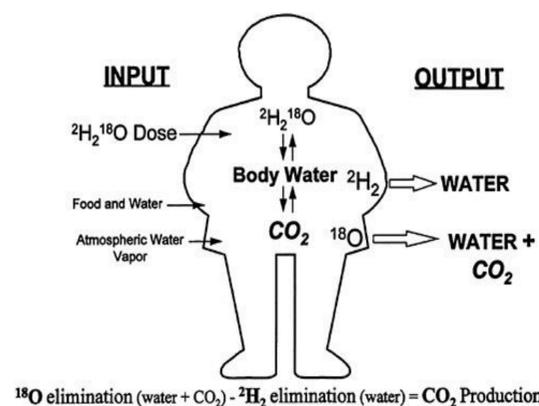
The influence of growth hormone on prepubertal children with Kabuki Syndrome is a novel field of research. Kabuki syndrome is a congenital anomaly/intellectual disability syndrome caused by a mutation in the *KMT2D* or *KDM6A* genes. These mutations causes distinct phenotypically features, such as short stature and facial dysmorphism. Earlier studies describe a high incidence of obesity in children with Kabuki syndrome. Studies in children with Prader-Willi syndrome, showed the effectiveness of growth hormone treatment on the fat-free mass^A, we expected the same effect in children with Kabuki syndrome. In this prospective study we investigated the influence of growth hormone on the total energy expenditure (TEE) and fat-free mass (FFM) in genetically proven, prepubertal children with Kabuki syndrome.



Material & Methods

We included 17 children with Kabuki syndrome, 8 girls and 9 boys with a mean age of 6.87 ± 2.23 year. The mean BMI SDS was 0.64 ± 1.76 .

The total body water (TBW) before start and during growth hormone treatment was measured with double-labeled water (DLW) technique. The DLW was also used to calculate the TEE in kJ/min. The FFM was calculated by dividing the TBW by the water percentage of FFM for children adjusted to age (Lohman et al. 1989)^B.



Results

The TEE was significantly increased by growth hormone from 3.60 ± 1.39 kJ/min to 5.51 ± 1.91 kJ/min ($p < 0.0001$). FFM was measured by dividing TBW by the water percentage of FFM. The FFM was significantly increased during growth hormone treatment compared to baseline (from 15.91 ± 6.43 kg to 17.10 ± 5.79 kg, $p = 0.014$).

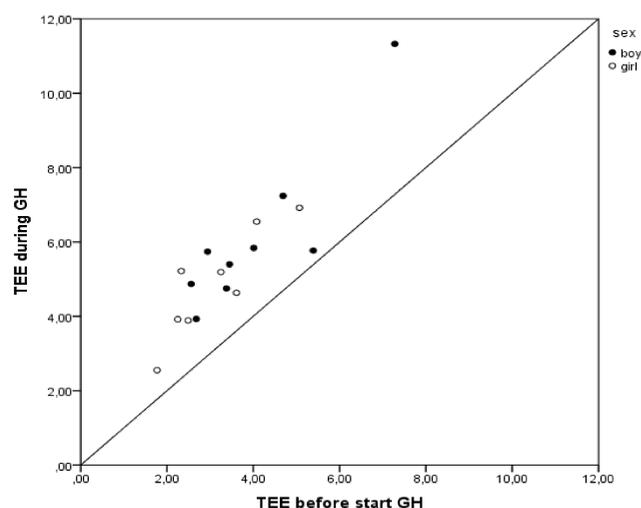


Figure 1: TEE significantly increased by growth hormone treatment.

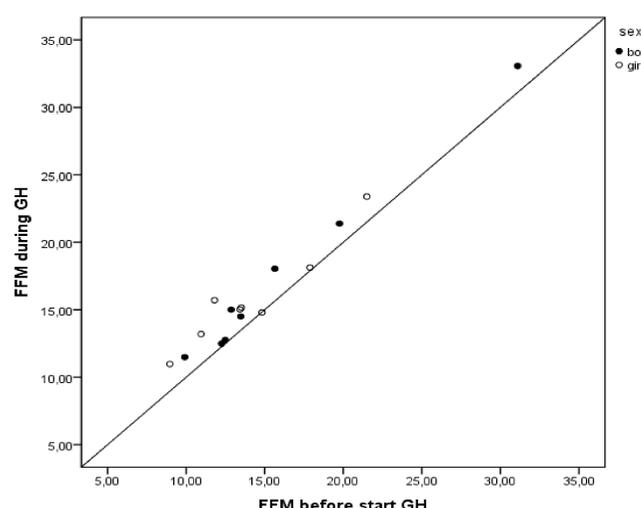


Figure 2: Growth hormone treatment significantly increased the FFM.

Conclusion

We investigated the effect of growth hormone treatment on the FFM in children with Kabuki syndrome. We found that the TEE and also the FFM increased significantly during growth hormone treatment compared to the baseline. We expect that the increase of metabolic activity and FFM reduce the chance to develop metabolic syndrome later in life.

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B. Lohman, T.G. Assessment of body composition in children. *Pediatr Exerc Sci*, 1. 1 (1989): 19-30.

