Reevaluation of congenital growth hormone deficiency in adulthood

**INTRODUCTION**

Congenital growth hormone deficiency (GHD) is a non-exceptional cause of short stature. The objective of our study is to reevaluate the clinical, biochemical, and evolutive features of congenital GHD in the region of Sfax in adulthood.

**PATIENTS AND METHODS**

This descriptive retrospective study included 48 patients over 16 years of old affected by GHD. It was conducted over 28 years (1990-2018) in our endocrinology department in Sfax, Tunisia.

**RESULTS**

- **Epidemiological and clinical features**:  
  - Our population is composed of 31 men and 17 women.  
  - Congenital GHD was revealed in 95.8% of cases by a short stature, noticed at a mean age of 9.4 ±3.8 years (2.25-15.5 years)  
  - **Family history**:  
    - consanguinity: 35.4%  
    - anterior-pituitary deficiency: 6.3%  
  - **Medical history**:  
    - neurosensory disorders: 3 cases  
    - malformations: 3 cases (digestive ++)  
  - **Birth history**:  
    - breech presentation: 13.8%  
    - intrauterine growth retardation: 12.1%  
    - foetal distress: 12.5%  
  - **Growth retardation was severe in 91.7% of cases** (height <3 standard deviation (SD) below the mean)  
  - **A delay in bone development was estimated at 3.7±2 years (1-12.25 years)**.  
- **Hormonal characteristics**:  
  
  **GH peak in stimulation tests**: 3.4 ± 2.8 ng/ml  
  
<table>
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<tr>
<th>GHD:</th>
<th>Total: 77%</th>
<th>Isolated: 60%</th>
<th>Partial: 23%</th>
<th>Combined: 40%</th>
<th>≥2 axis: 42%</th>
<th>(gonadotropic +)</th>
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- **Radiological characteristics**:  
  - Pituitary MRI:  
    - Abnormal: 60.4%  
    - anterior pituitary hypoplasia: 86.2%  
    - pituitary stalk defects: 44.8%  
    - ectopia of posterior pituitary: 34.5%  

- **Therapeutic and evolutive characteristics**:  
  - The majority of our patients (91.7%) received recombinant human growth hormone (GH).  
  - **At the start of GH treatment (TTT)**:  
    - CA = 11.3 ± 3.2 years (4.75-19 years)  
    - BA = 6.9 ± 7.8 years (3 months-13 years)  
    - Weight = 22.4 ± 8.8 Kg (11-53 kg)  
    - Height = 115 ± 16.2 cm (78-152 cm)  
  - Our patients were divided into 3 groups:  
    - **Ceasing GH TTT: 9/44 (20.5%)**  
      - Having problems in TTT cover/lost to follow up  
    - **Still under GH: 6/44 (13.6%)**  
    - **TTT duration**: 73.5 ± 32.6 months (48-120 months)  
    - Height gain: 1.8 ± 2.1 DS (0-5 DS)  
    - Height ≤2 DS: 4/6 cases  
    - Mean: 2/6 cases  
    - ≥2 DS: 0/6 cases  
    - **TTT was stopped as the bone age ≥13 ′-15 ′ years**:  
      - 29/44 (65.9%)  
    - GH dose: 0.64±0.10 IU/kg/week  
    - TTT duration: 55.9 ± 30 (12-120 months)  
    - Height gain: 1.8 ± 1 SD (-1, + 3 DS)  
    - Target height reached: 3/20 cas (15%)  
  - 72.4% of patients who received GH did hit puberty.

- **Hormonal revaluation of patients receiving/ed GH: n=44**  
  - Same hormonal status: 61.4%  
  - Detection of another hormonal deficiency: 13.6%:  
    - corticotropic: 4  
    - thyreotropic: 3  
    - gonadotropic: 2  
  - Restoration of the somatotropic axis: 1 patient with an isolated GHD and a normal MRI  
  - Non-available: 22.7%  

**CONCLUSION**

Although it is a rare condition, missing the diagnosis of DGH will result in poor growth and short stature adults. GHD may or may not persist into adult life and associate or not with other hormonal deficiencies. Patients with childhood onset GHD are usually retested in late adolescence or young adulthood thus the importance of a close collaboration between the paediatric and adult endocrinologists during the transition period.