Sleep Medicine 66 (2020) 21–23

Contents lists available at ScienceDirect

Sleep Medicine

journal homepage: www.elsevier.com/locate/sleep

Images in Sleep Medicine

Obstructive sleep apnea in Hutchinson-Gilford progeria

Bertien Buyse [a, b, *], David Cassiman [a, c], Dries Testelmans [a, b]

[a] KU Leuven, Department of Chronic Disease, Metabolism and Ageing, Leuven, Belgium
[b] University Hospital Leuven, Leuven University Centre for Sleep and Wake Disorders and Department of Pulmonology, Leuven, Belgium
[c] University Hospital Leuven, Department of Hepatology, Leuven, Belgium

ARTICLE INFO

Article history:
Received 3 June 2019
Received in revised form
29 July 2019
Accepted 1 August 2019
Available online 8 August 2019

Keywords:
Obstructive sleep apnea
Retrognathia
Progeria
Cardiovascular abnormalities
Hypersomnolence
Fatigue

1. Introduction to the case

The Hutchinson-Gilford progeria syndrome (HGPS) is an extremely rare genetic disease (prevalence 1/20.000.000), characterized by premature aging. The disease is related to a defect in the lamin A/C gene with deleterious effects on cell division. The patients develop rapidly progressive atherosclerosis, generally leading to death from myocardial infarction or stroke at the age of 13–15 years [1,2]. We present a case of a HGPS patient with a diagnosis of moderate obstructive sleep apnea (OSA) treated with continuous positive airway pressure (CPAP).

2. Image analysis

The patient was a 19-year old male diagnosed with HGPS. He complained of fatigue and lack of energy. His Epworth Sleepiness Scale (ESS) score was 13/24. He was working as a salesperson in a computer store; yet, due to high sleep need he could only work during the afternoon hours. He took experimental drugs under supervision of the Boston Children's Hospital in the United States: lonafarnib in combination with pravastatin and everolimus. His height was 128 cm and his weight was 21 kg (body mass index: 12.8 kg/m^2) and typical clinical signs of progeria such as alopecia, a giant size appearance of the skull in contrast to the body and face, loss of subcutaneous fat and dental crowding due to limited size of the mandible were present (Fig. 1).

Recently, his father, who was coincidentally staying in the same hotel room, observed snoring and apnea during sleep.

An attended polysomnography was performed and demonstrated a sleep efficiency of 92% with an increased arousal index of 36 per hour of sleep. The apnea-hypopnea index (AHI) was 26 per hour of sleep, and was higher during rapid-eye movement (REM) sleep (AHI in REM 75/h, in non-REM 20/h) with deep oxygen desaturations (Fig. 2). There was no overt positional modulation: AHI in non-REM supine 20/h versus 18/h in non-supine, AHI in REM supine 81/h versus 72/h in non-supine, see also Fig. 2. The oxygen desaturation index (ODI, 3%) was 23/h. The events were obstructive (obstructive apnea index 12/h and obstructive hypopnea index 14/h). Manual in-lab CPAP titration under polysomnography was performed: CPAP treatment with a pressure of 8 cm H2O led to normalization of the AHI (1/h) (Fig. 2).

The patient was re-evaluated two months later. He used his CPAP-device every night for more than 7 h. His ESS score dropped to 3/24. His sleep need was reduced (8 instead of 10 h) and he was able to work also during the morning hours. At one-year follow-up, he was still very adherent to his CPAP treatment.

3. Discussion

Progeria patients generally demonstrate an impressive craniofacial disproportion in the form of a large skull in contrast to a small face with obvious micrognathia [1,3]. However, to the best of our knowledge, OSA has never been reported. We speculate that the presence of OSA might be underestimated in HGPS patients, because they remain small and are extremely underweight. Moreover, the fatigue complaint, as present in our patient, is easily attributed to the aging process. OSA in progeria can be treated with CPAP with positive impact on fatigue, sleepiness and quality of life. The impact of CPAP treatment on the very progressive cardiovascular aging process remains an open question.

* Corresponding author. University Hospital Gasthuisberg, Herestraat 49, 3000, Leuven, Belgium.
E-mail addresses: Bertien.Buyse@uzleuven.be (B. Buyse), David.Cassiman@uzleuven.be (D. Cassiman), Dries.Testelmans@uzleuven.be (D. Testelmans).

https://doi.org/10.1016/j.sleep.2019.08.001

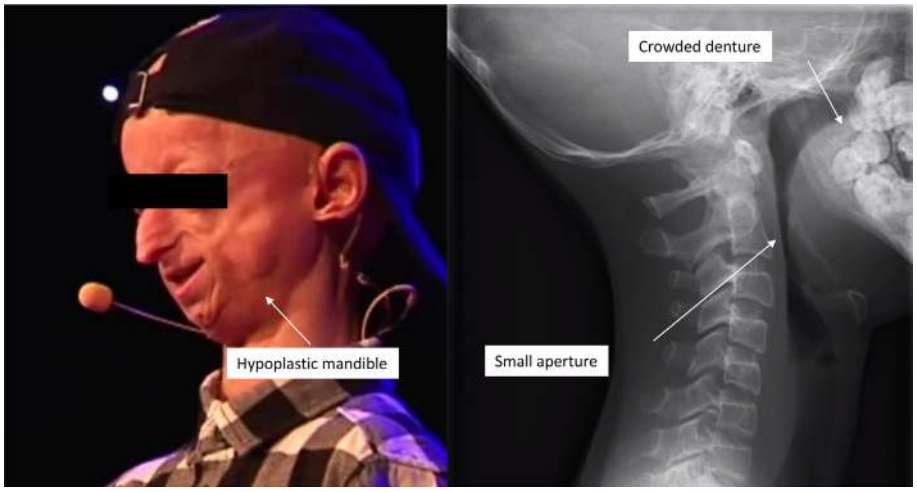

Fig. 1. Profile picture of the patient and profile x-ray of the cervical spine, tongue base and denture.

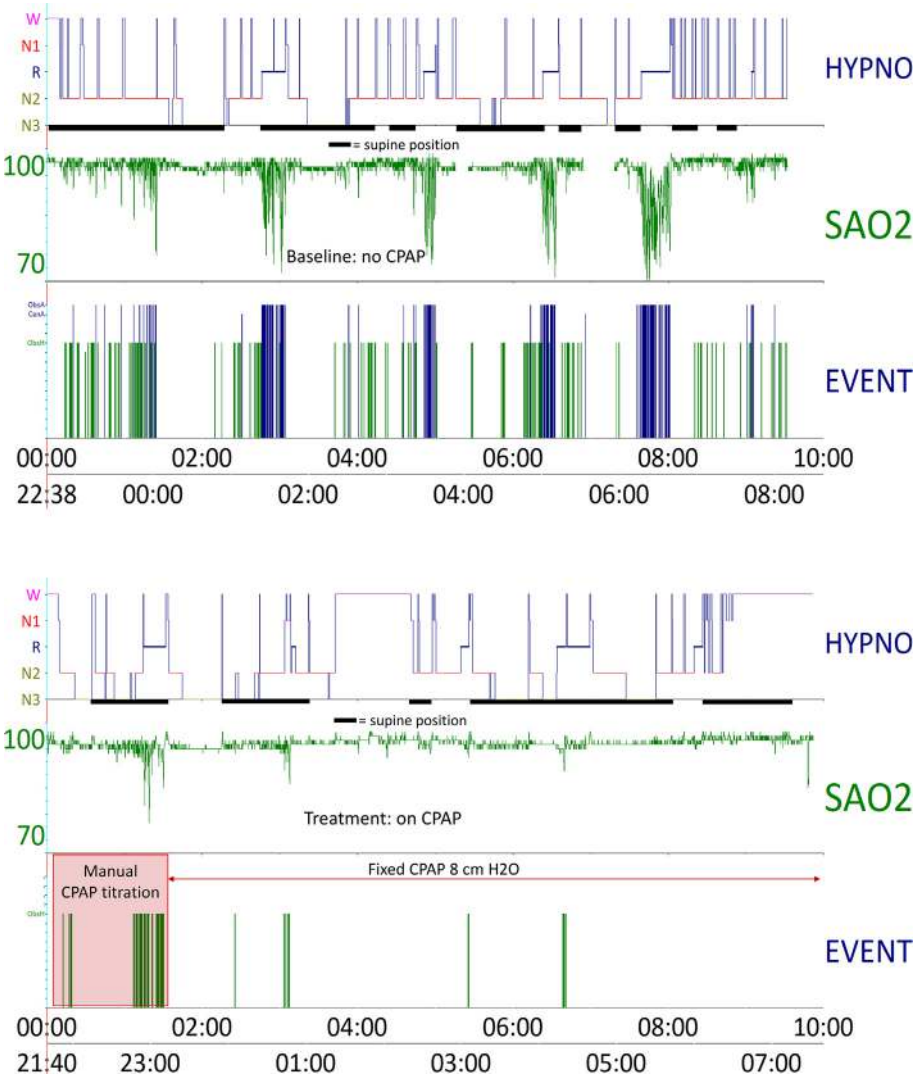

Fig. 2. Hypnogram (hypno), oxygen saturation (SAO2) and respiratory events (event) on the diagnostic night (upper part) and therapeutic night (lower part); W = wake, N1 = stade 1, N2 = stade 2, N3 = stade 3, R = rapid eye movement sleep.

Institution at which work was carried out

KU Leuven/Leuven University Centre for Sleep and wake disorders – University Hospital Leuven, Belgium.

Financial support

No financial support was obtained for this case report.

Consent

Informed consent was provided and the study was approved by the ethical commit-tee of the University Hospitals Leuven (approval number B322201213466).

Conflict of interest

No conflict of interest.

The ICMJE Uniform Disclosure Form for Potential Conflicts of Interest associated with this article can be viewed by clicking on the following link: https://doi.org/10.1016/j.sleep.2019.08.001.

References

[1] Ahmed MC, Ikram S, Bibi N, et al. Hutchinson-gilford progeria syndrome: a premature aging disease. Mol Neurobiol 2018;55:4417–27.
[2] Gordon LB, Shappell H, Maasaro J, et al. JAMA 2018;3319(16):1687–95.
[3] Chawla GS, Agrawal PM, Dhok A. Progeria: an extremely unusual disorder. Skeletal Radiol 2017;46:1149–53.