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STUDIES OF SLEEP, MRI AND MR-SPECTROSCOPY IN AN ADULT PATIENT WITH PRADER-WILLI-SYNDROME

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The Prader-Willi-Syndrome is characterized by infantile muscle hypotonia, hypogonadism with genital hypoplasia, mental deficiency, childhood-onset malignant obesity and characteristic skeletal dysmorphology. Despite a Broca-Index of 155% and other clinical features predisposing for obstructive sleep apnea the main finding in this adult patient with Prader-Willi-Syndrome were the irregular breathing pattern and recurrent hypoxemia during sleep. During the day the oxygen saturation showed only minimal fluctuations, whereas during the sleep oxygen fluctuations of more than 10% were registered. For further work-up the patient recieved a mobile multisensor screening device (MESAM III) which deteced cardiac arrhythmia and snoring during sleep. Two nights of sleep polygraphy were registrated in our laboratory. During this nights snoring was present, but without pathological apneas (less than 10 sec duration). More prominent was the finding of an irregular breathing pattern with almost constant changes between hypoventilation and hyperventilation, accompanied by diminuition of oxygen saturation of more than 10%. This finding was especially marked during phases of REM sleep.
During spontaneous sleep Magnetic Resonance Imaging and localized IH-Spectroscopy were performed at 1.5 Tesla. Oxygen saturation was monitored by pulsoxymetry. Sagital T1 weighted images revealed a widening of the 4th ventricle and a diminished a.p. diameter of the lower brain stem. Despite of peripheral oxygen fluctuations between 98% and 85% localized IH-Spectroscopy did not show any evidence of lactate .

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