Myeloid leukemia and Prader-Willi syndrome – A case report

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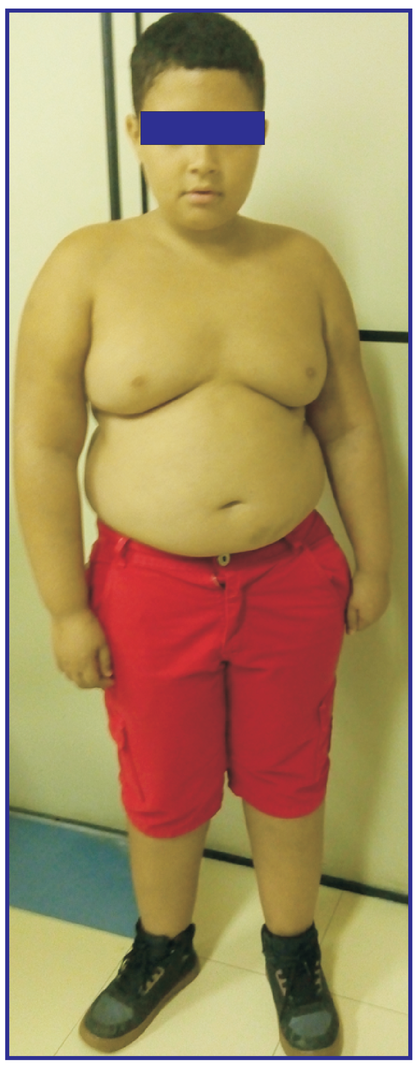
CASE REPORTS

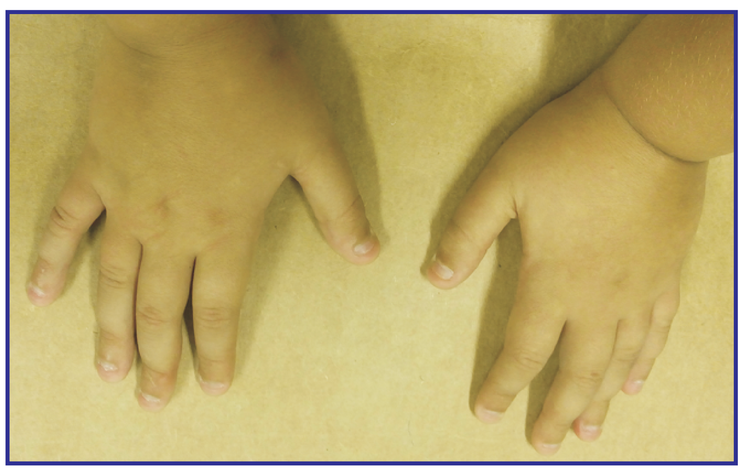
A 6-year-old boy was referred to INCA's Clinical Genetics because of dysmorphic features. He had acute leukemia at 4 years-old. Clinical exam demonstrated mental retardation, short stature, obesity, unilateral chryptorchidism, and small hands and feet. Hypotonia at birth, hyperphagia, compulsive nails picking, and food-seeking behaviors were noted.

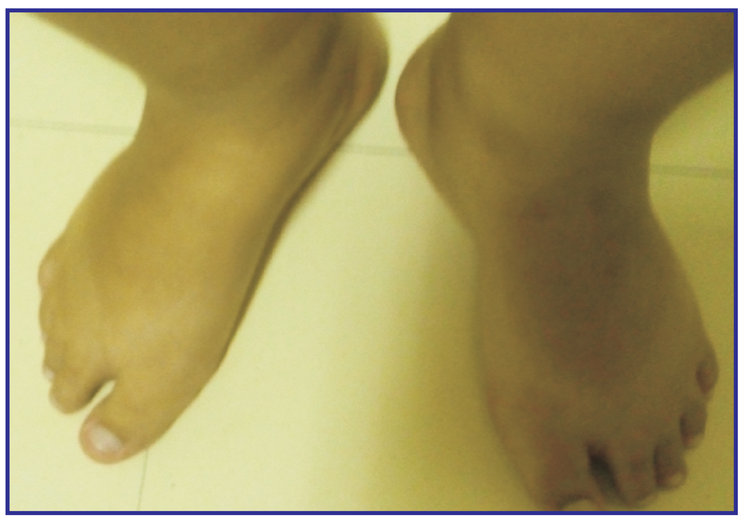
METHODS

The DNA methylation analysis at 6' SNRP (Small Nuclear Ribonucleoprotein-associated Protein N) gene was abnormal.

MLPA (Multiplex Ligantion Probe Amplification) - deletion of 15q11-13.







DISCUSSION AND CONCLUSIONS

Prader-Willi syndrome (PWS) is characterized by severe hypotonia and feeding difficulties in early infancy, followed in later infancy or early childhood by eating and gradual development of morbid obesity. Motor milestones and language are delayed. All individuals have some degree of cognitive impairment. This patient had clinical criteria for disease. The DNA methylation analysis at 6' SNRP (*Small Nuclear Ribonucleoprotein-associated Protein N)* gene was abnormal. This test can identify 97% of cases, but can't identify the etiologic class (15q11-13 deletion, abnormal parent –specific imprinting on 15 or uniparental disomy). MLPA (*Multiplex Ligantion Probe Amplification*) revealed deletion of 15q11-13. Torrado et al (2007) studying 91 individuals with a deletion showed a higher frequency of need for special feeding techniques, sleep disturbance, and speech articulation defects. Our patient presents all this characteristics. There are few reports of cancers among persons with PWS. Patja e cols identified 56 PWS patients from the Finnish registry. There were three cancers (acute lymphatic leukemia, testicular cancer and breast cancer) during the follow up; the expected number based on average Finnish population was 1.51 (SIR 2.0, CI95% 0.4-5.8). Daves e cols (2003), published a coorte of 1160 persons with PWS and observed three cases of myeloid leukemia vs 0,075 leukemias expected (p=0,0001).

There is a probably increased risk of cancer among persons with PWS. Myeloid leukemia must be thought in PWS patients.

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Meeting

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