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Early-onset type 1 diabetes and multiorgan autoimmunity in a girl with partial monosomy 2q and trisomy 10p

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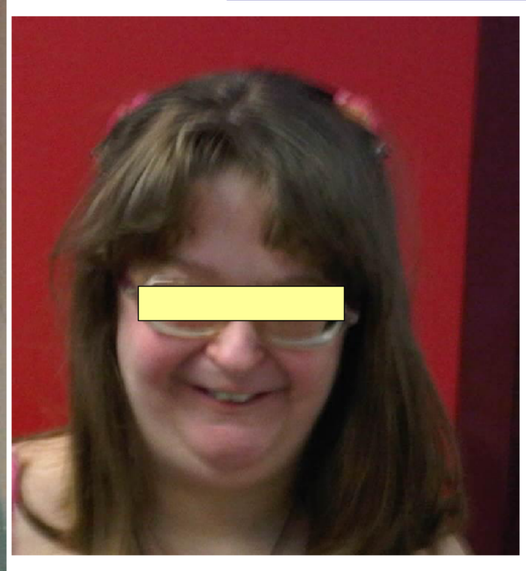
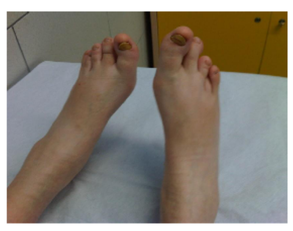
OBJECTIVES

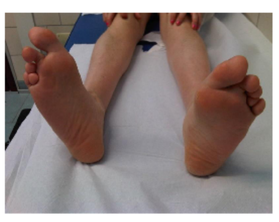
Genes in the human leucocyte antigen (HLA) region confer about 50% of the genetic risk of T1DM. More than 40 different genes give a minor contribution to T1DM risk, some of them are related to the immune function.

Case presentation

- A girl was referred at the age of **9 months** with severe ketoacidosis in T1DM at onset. Anti-islet cells and anti-insulin autoantibodies were negative at diagnosis, as well as screening for neonatal diabetes and methylation analysis for Prader-Willi syndrome. She was the only daughter of unrelated Caucasian parents, born at term by vaginal delivery. The father was affected by Crohn disease. **She showed round face, prominent forehead, upslanting palpebral fissures, deep-set eyes, midface hypoplasia, and depressed nasal bridge. Mild mental retardation, hypotonia and eczema were evident.** At the age of **3 years** she developed juvenile idiopathic arthritis. Hypertriglyceridemia and anti-thyroperoxidase, anti-thyroglobulin autoantibodies were first detected at the age of **16 years**, but thyroid function remained normal over time. She manifested growth retardation and pubertal delay with low bone mineral density and 3 fractures from mild trauma. Spontaneous menarche occurred at the age of **17 years**. Final height was 154 cm (-1.7 SDS), significantly lower than mid-parental height (cm 165, 0.2 SDS). Recurrent seizures first appeared at the age of **16 years**.







RESULTS

CGH-array analysis: complex rearrangement involving chromosome 2q deletion and chromosome 10p duplication [2q37.3 (238.525.260-243.041.364) x1, 10p15.3p14 (148.206-6.633.649) x3]

CONCLUSIONS

Analysis of 2q and 10p regions revealed that **PDCD1** (programmed cell death 1 precursor) gene is located on chromosome 2, **IL-2RA** (interleukin 2 receptor, alpha chain precursor/CD25) gene is located on chromosome 10. They are involved in the regulation of T cell function during immunity and tolerance. Their duplication or deletion could be responsible for changes in T regulatory cells affecting their ability to suppress effector T cell function, finally increasing susceptibility for autoimmune diseases. A previous paper (published on Feb 2013) described the same complex rearrangement in a patient affected by Turner syndrome, T1DM and Hashimoto's thyroiditis

References

Grossi A et al. Multiorgan autoimmunity in a Turner syndrome patient with partial monosomy 2q and trisomy 10p. Gene. 2013; 515(:439-43.

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