A case of a family with van der Woude syndrome
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Objective
Van der Woude syndrome is an autosomal dominant syndrome that affects the development of the face. The cause is mutation in the IRF6 gene, which is located in the long arm of chromosome 1 (1q32. 3-q4). The gene provides instructions for making a protein that plays an important role in early development. The protein is active in cells that give rise to tissues in head, face, skin and genitals. Children with VW syndrome are born with a cleft lip of palate, depressions (pits) near the center of the lower lip, which is usually moist, due to the presence of salivary and mucous glands in the pits. Hypodontia (absent teeth) is the most common anomaly. They also have an increased risk of delayed language development, learning disabilities and other mild cognitive problems, but IQ is usually within normal ranges.

Methods
In our family, many clinical conditions due to the VW syndrome were identified. Grandfather had pits on the lower lip, troubles with salivary gland, and speaking problems due to the healed up tongue. (My/His?) daughter had pits on the lower lip, troubles with salivary gland as well, hypodontia, and 4 missing teeth. Her first born child (born in 2011) had congenital palatoschisis, pits on the lower lip and troubles speaking. Her second pregnancy in the year 2013 ended tragically, with death of foetus at 36 weeks due to preeclampsia and placental abruption. Her third pregnancy ended with a c section at 32 weeks because of preeclampsia. The new-born had cleft upper and lower lips, and pits on the lower lip (picture). So far she is developing fine.

Results
Genetic tests were made in all previously described individuals. Mutations in gene IRF6 (16C>T), van der Woude syndrome was confirmed in all 4 of them.

Conclusion
Van der Woude syndrome is an autosomal dominant syndrome, mutation in gene IRF6 is present. It is clinically associated with cleft lips and palate, pits on lower lips and palate, salivary gland and speaking problems, missing teeth, learning disabilities and other mild cognitive problems. In our family, mutation in gene IRF6 was diagnosed with genetic testing, clinically 4 members had clinical signs described above. Face development anomalies can be diagnosed early, in utero with ultrasound. When a child is at least 2-3 months old, oral and maxillofacial surgeries can fix palate, lips and dental anomalies. Today, the prognosis and treatment of individuals with VW syndrome is excellent.