17th World Congress in Fetal Medicine

Family with Van der Woude syndrome

Predič M, Mujezinović F, Šurc A
Štorklja d. o. o, Ljubljana, Slovenia

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, that is located in the long arm of chromosome 1 (1q32. 3-q4). Gene provides instructions for making a protein that plays an important role in early development. Protein is active in cells that give rise to tissues in head, face, skin and genitals. People 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 withing normal ranges.

Methods

In our familly many clinical conditions due to the VW syndrome were identified. Grandfather had pits on lower lip and troubles with salivary gland and speaking problems due to healed up tounge. Daugther had pits on lower lip, also troubles with salivary gland and hypodontia, 4 missing teeth. Her first born child (born in 2011) had congenital palatoschisis and pits on lower lip and troubles speaking. Second pregnancy in the year 2013 ended tragically with death of foetus in 36. week due to preeclampsia and placentar abruption. Third pregnancy ended with c section in 32. week, because of preeclampsia. Newborn had cleft upper and lower lip and pits on 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 gene is present. It is clinically associated with clift lips and palate, pits on lower lips and palate, salivary gland and speeking 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 child is at least 2-3 months old oral and maxillofacial surgeons operate to fix palate, lips and dental anomalies. Prognosis and development of individuals with VW syndrome today is excellent.