Presented review of literature data (medline, eLIBRARY.RU, PabMed) identifies typical problems of parents whose children were born with cleft lip and palate (RGN), as well as research directions in improving the organization of medical care for parents and patients with UAH in solving these problems. Analysis of domestic and foreign authors on MEDLINE databases, eLIBRARY.RU, PabMed Still lacks research on the experiences of parents with RSN, what typical problems they encountered in the maternity hospital and, especially, at home. It is now necessary to carry out research that has traditionally been rely not only on psychological approaches, but also on broader prospects for research in the field sociology, social policy, nursing and health care using both qualitative and quantitative methods. In addition, there is a lack of research in the field of cleft lip and palate to study the experience and needs of parents at different stages of their children’s lives.

93 WILLIAMS-BEUREN SYNDROME: A CASE REPORTS
Dorian Laslo*, Vlilja Tomac, Silvija Pušeljić. Faculty of medicine Osijek, Josip Juraj Strossmayer University of Osijek
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Introduction Williams-Beuren syndrome (WBS) is developmental disorder caused by microdeletion of genes from chromosome 7. It is estimated in 1 of 10 000 people. WBS could be autosomal-dominant inherited, but usually, it is caused by de novo microdeletion of 26-28 genes on chromosome 7. Usually genes such as CLIP2, NCF1, ELN, GTF2I, GTF2IRD1 and LIMK1 are deleted. ELN gene is detected as a cause of supra-valvular aortic stenosis, while absence of NCF1 gene is related to hypertension. Patients with WBS are characterized by cardiovascular diseases, facial dysmorphic features, intellectual disability, unique personality character and endocrine abnormalities.

Case Report We present the case of three patients, two girls at the ages of one and three years and a boy at the age of two years. All three patients have healthy parents and brothers and sisters without known chronic or genetic diseases. All three were presented with facial dysmorphic features which include broad nasal bridge, microretrognathia, large mouth, upper lip is thin while lower lip is thicker. All three patients are cognitively deficient and they have speech problems. The female patient at the age of three years also has hypertension, small teeth, larger neurocranium, atrial and ventricular (muscular) septal defect, gastroesophageal reflux disease (GERD), hypothyroidism and endocrine disorders; hypocalcemia, hypercalciuria, hypothyroidism and early puberty. The male patient also has prominent ears, palmar crease on left palm, pulmonary stenosis, atrial septal defect, gastroesophageal reflux disease, hypothyroidism and early puberty. The male patient also has speech problems. Language skills represented as vocalizations. Physical growth of our patients fluctuated from 3 to 75 percentiles for height and weight. The other clinical features include hypotonia and strabismus (all patients), autistic behavior (2 patients), ataxia, seizures, hepatomegaly (one in each patient). Dysmorphic features were non-specific.

Ophthalmological examination revealed congenital high myopia (2 patients), partial atrophy of an optic nerve (2 patient), nystagmus (1 patient), astigmatism (1 patient), retinal atrophy (1 patient).

Biochemical analysis showed elevated creatine kinase from 1874 to 6266 (averaging 3754 U/L), ALT from 42 to 93 (68 U/L), AST from 53 to 106 (80 U/L), LDH from 370 to 503 (437 U/L).

Electromyographic examination has shown that all children had signs of primary muscle lesion. Muscle MRI has displayed a severe atrophy muscles and fatty infiltration in one patient. MRI findings have reported pachygria and ventriculomegaly (all patients), hypoplasia cerebellum, corpus callosum, pons (2 patients), hypoplasia of temporal lobes (1 patients), cerebellum cysts (2 patients). EEG did not reveal epileptic form activity.

We revealed compound heterozygous mutations in all three patients. These were five different mutations: missense c.385C>T (p.R129W) and c.1325G>A (p.R442H), nonsense