10th International Conference on

Otolarngology: ENT Surgery

May 15, 2023 | Webinar

Received Date: 04-03-2023| Accepted Date: 07-03-2023 | Publication Date: 30-05-2023

A rare case of hearing loss at a Patient with "Czech Dysplasia"

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"Czech dysplasia" is an inherited condition that affects joint function and development. It is caused by a particular mutation in the COL2A1 gene which provides instructions for making a protein that forms type II collagen. It is characterized by early-onset, progressive pseudo-rheumatoid arthritis and hypoplasia/dysplasia of the third and fourth metatarsals. Some people have progressive sensorineural hearing loss. This is an extremely rare disease, with only 28 cases described in the medical literature so far.

Case report: Women (55) present with rapidly progressive hearing loss. In her medical history, she is a typical orthopedic patient for many years with both of her hips and knees have been operated on and replaced with a prosthesis. Genetic testing was performed and "Czech dysplasia" was confirmed. Due to the complications of her illness, she has been receiving a lot of antibiotics, including ototoxic ones. Her otoscopic findings were normal. Audiometry testing showed bilateral moderate-severe hearing loss. Hearing aids were advised and the patient was very satisfied with her hearing.

The only dilemma remains whether her hearing loss is a consequence of many ototoxic drugs she has received or is a part of the underlying disease.

Biography

Ana Krivokapic is an ENT specialist, working in primary health care in Belgrade. She has been an active participant in many national conferences and symposiums in Serbia, and international ones in Brussels, Milano, Vienna, etc

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