Titel des Beitrags: Non-syndromal round window atresia: an autosomal dominant genetic disorder with variable penetrance?

Abstract: Round window atresia can be seen in association with syndromal anomalies such as mandibulofacial dysostosis, Mondini type anomalies or cretinism, or with extensive otosclerosis. Non-syndromal round window atresia is extremely rare and until today there is no proof whether non-syndromal round window atresia can be inherited. We present two family members with non-syndromal round window atresia. Based on this case and on an extensive literature review, we discuss the possible ways of sound transmission with round window occlusion as well as the possibility of an autosomal dominant inheritance pattern of these two non-syndromal cases. Evaluating the hearing test results, the effect of round window atresia in non-syndromal cases cannot be thoroughly explained by current theories of sound transmission. In general, full occlusion of the round window should result in a complete air-bone-gap and a surgical procedure to open the window, e.g. a cochlear fenestration which creates a new pressure outlet for the inner ear fluids, should result in normalizing the hearing levels. Astonishingly, patients with non-syndromal round window atresia, show hearing tests similar to those in patients with otosclerosis. In contrast to otosclerosis, complete closure of the air-bone-gap by surgery in patients with round window atresia does not seem to be possible according to currently reported cases. Therefore, routine examination of the round window in stapes surgery should be part of every stapes surgery.
and a high resolution CT scan should be considered in particular prior to revision of unsuccessful stapes surgery. As a genetic inheritance pattern can be assumed, specific emphasis should also be laid on the patient's family history.

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