Erfolgreiche Hörrehabilitation einer synaptischen auditorischen Neuropathie mittels Cochlea Implantat

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Einleitung

• 72-jährige Patientin mit einer **beidseitigen synaptischen auditorischen Neuropathie**

• Beidseitige progrediente Hörminderung mit ausgeprägtem Diskriminationsverlust

• Stabiles Tonaudiogramm - Abfall der Sprachverständlichkeit beidseits mit stärkerer Ausprägung rechts

Methoden

• **Tonaudiometrisch** seitensymmetrische milde Presbyakusis

• **DPOAE** -Korrelation zum Tonaudiogramm

• **Sprachaudiometrie**-korrelierenden Hörverlust für Zahlen bei deutlichem Diskriminationsverlust mit ausgeprägter Regressionskurve beidseits

•Die **Aggravationsprüfungen** fielen negativ aus

•Ausfall der **Stapediusreflexe**

• Pathologischen Schwellenschwund im **Carhart-Test** und herauslaufende Mithörschwelle in der **Geräuschaudiometrie nach Langenbeck**

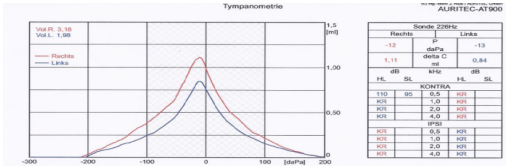
• Die wiederholt durchgeführte **BERA** zeigte nur noch links bei 100dB eine deutlich verlängerte Welle V

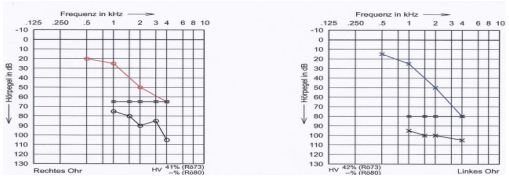
• Bei **fehlender Welle I** trotz unauffälliger DPOAE- V.a. synaptischer auditorischen Neuropathie

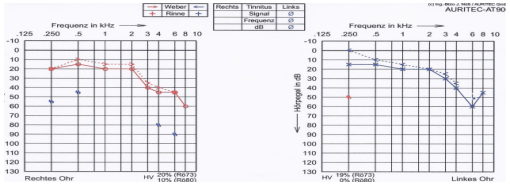
• **Transtympanale Elektrocochleografie** rechts- Bei regelrechten Cochlea Microfonics kein Aktionspotential

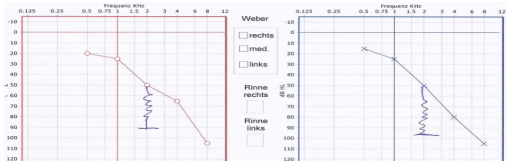
• Best aided **Hörgerätversorgung**- Sprachverstehen von 50% links und 0% rechts =nicht suffizient

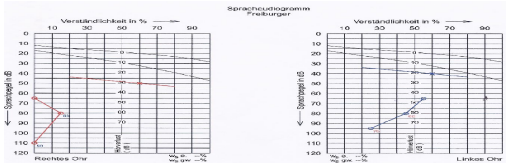
• Bei funktioneller Taubheit rechts erfolgte zunächst die **Cochlea Implantation** rechts

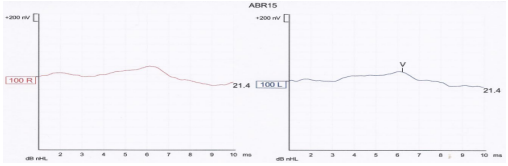


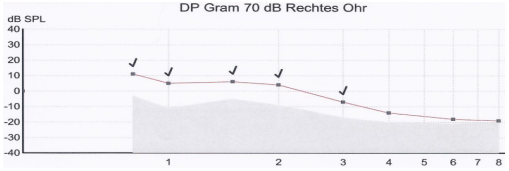


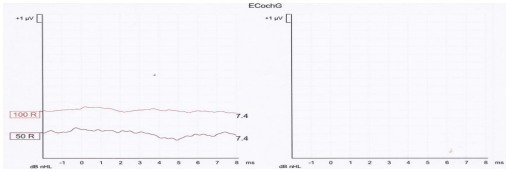












Ergebnisse

• Intraoperativ konnten regelrechte **Nervenantworten** gemessen werden

• Die elektrisch evozierte und optisch registrierte **Stapediusreflexschwelle** zeigte keine außergewöhnlichen Abweichungen von den gemessenen Nervenantworten im Sinne einer Reizweiterleitungsstörung.

• Die Ergebnisse schon bei der Erstanpassung zeigten eine **regelrechte Nervenstimulation** und Weiterleitung des Signals

Schlussfolgerung

• Die **aktuelle Datenlage** ist aufgrund der geringen Fallzahlen nicht ausreichend dokumentiert

• Oft ist die **Versorgung mit einem Cochlea Implantat** die einzige Möglichkeit ein Sprachverstehen wiederzuerlangen

• **Postoperative Performance** der Patienten -sehr breit gefächert - erreicht nicht den durchschnittlichen Hörgewinn der großen Patientengruppe mit cochleärer Ertaubung

• Die **Prognose** im Fall einer **isolierten synaptischen Neuropathie** im Bereich der Cochlea erscheint günstiger und vergleichbar mit dem Patientengut der Innenohrertaubungen

• Bei einer auditorischen Neuropathie -**realistische Beratung** des Patienten

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Five months prior aspiration cytology of this nodule revealed no malignant cells. 99 mTc thyroid scan demonstrated hyperfunctional areas in the middle and lower part of each lobe. The screening of mutations of the RET- protooncogene performed for exon 10, 11, 13, 14, 15, and 16 detected an extremely rare mutation involving codon 649 (replacement of a serine by a leucin, TCG>TTG) in exon 11, present in a heterozygous state. Genetic testing of all four first degree relatives (two daughters: 40, 42 yrs. and two grandchildren: 8, 14 yrs.) determined all of them as mutation carriers. Surgery was recommended.
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The thyroid specimen measured 45 x 12 x 80 mm, 23 g. On cut section a grayish-brown encapsulated follicular adenoma, measuring 14 mm in diameter was present in the middle portion of the left lobe. Microscopically adjacent to this mass a 4 x 2 mm well-circum-scribed, nonencapsulated neoplasia, arranged in solid and follicular growth patterns, containing colloid material in the lumen, was found. The spindle tumour cells showed moderately partially stronger atypia.
The nuclei appeared enlarged and hyperchromatic. There were amorphous eosinophilic amyloid depositions within the tumour. Fig. 1 d-e) No signs of lymph or blood vessel invasion were found. Immunohistochemistry was positive for calcitonin, chromogranin, and thyroglobulin. Thus histopathological andimmunohisto-chemical findings showed this to be a mixed medullary-folli-cular microcarcinoma of the thyroid. Fig. 1 f-h) No metastatic foci were present. Tumour staging according to the International Union against Cancer (TNM) classification was pT1 pNx pM0 R0 L0 V0. Postoperative course was uneventful. Two months after surgery basal serum calcitonin level had fallen to 5,2 ng/l (normal: < 10 ng/l). The patient had normal results of serum calcium, intact PTH, 24-h urinary catecholamines, and plasma free metanephrines, respectively. Because of the component with follicular features of the thyroid carcinoma she received radioiodine irradiation in February 2006. A therapeutic dose of 3.85 GBq 131I was administered. In October 2006 total body scan with 387 MBq 131I revealed no radioactive iodine uptake in the thyroid bed, bones, or lungs. Repeated measurements of basal serum calcitonin levels were normal, subsequent thyroglobulin concentrations were undetectable (< 0.3 µg/l) under replacement therapy. To date there has been no evidence of recurrent or metastatic disease. All four first-degree relatives (two daughters and their two children) were tested for basal and pentagastrin stimulated calcitonin levels and underwent ultrasound examination of the thyroid. These results were in normal range and ultra-sonography confirmed a normal thyroid gland in all cases. After extensive discussion regarding the documented genetic transmission three individuals agreed to surgery.
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The two daughters of the index patient were both prophylactically thyroidectomized in March 2007. Histopathologic and immunohistochemical findings showed nodular hyperplastic CCH (0.1 cm in diameter) plus focal CCH in one case and only focal CCH in the other. No premalignant stages of MTC. Fig. 1 a-c) The 14 year old grandchild of the male patient also underwent prophylactic thyroidectomy in July 2007. At this area of 6 y a man with a slightly increased number of C-cells was found, but no CCH. To date the parents of the other 8 year old grandchild declined this prophylactic procedure. An at least annual monitoring of basal and pentagastrin stimulated calcitonin levels was offered (last in April 2008 without pathological increase) and preventive thyroidectomy as soon as these become abnormal.

HELIOS Kliniken
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Whatever the kind of RET mutation is, optimal treatment in gene carriers should take into account the potential aggressiveness of the disease and the fact, that histopa-thologic stage is the main prognostic factor of MTC. There fore once gene carrier status is established, the need to perform prophylactic thyroidectomy is undisputed, but, especially in cases with rarely reported RET mutations, the appropriate timing is unclear. Our case indicates that the RET Ser649Leu mutation is associa-ted with late-on set non-aggressive disease. Therefore recommendations for surgery should be individualized depending on basal and stimulated calcitonin levels.

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Five months prior aspiration cytology of this nodule revealed no malignant cells. 99 mTc thyroid scan demonstrated hyperfunctional areas in the middle and lower part of each lobe. The screening of mutations of the RET- protooncogene performed for exon 10, 11, 13, 14, 15, and 16 detected an extremely rare mutation involving codon 649 (replacement of a serine by a leucin, TCG>TTG) in exon 11, present in a heterozygous state. Genetic testing of all four first degree relatives (two daughters: 40, 42 yrs. and two grandchildren: 8, 14 yrs.) determined all of them as mutation carriers. Surgery was recommended.
The 67 year old index patient underwent total thyroidectomy without lymph node dissection in August 2005. Intraoperatively there was no evidence of MTC or cervical lymphaden-opathy.
The thyroid specimen measured 45 x 12 x 80 mm, 23 g. On cut section a grayish-brown encapsulated follicular adenoma, measuring 14 mm in diameter was present in the middle portion of the left lobe. Microscopically adjacent to this mass a 4 x 2 mm well-circum-scribed, nonencapsulated neoplasia, arranged in solid and follicular growth patterns, containing colloid material in the lumen, was found. The spindle tumour cells showed moderately partially stronger atypia.
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