**Supplementary information, Data S1 Materials and Methods**

***Clinical Evaluations***

The DDOD pedigrees 1 and 3 were ascertained from Shanxi province, and pedigree 2 was from Jilin province of mainland China. All the procedures were approved by the Ethics Reviewing Committee of the Chinese PLA General Hospital and were carried out only after written informed consent was obtained from each individual and/or parents of the children. The medical history was obtained by use of a questionnaire regarding the following aspects: age at onset, evolution, symmetry of the hearing impairment, hearing aids or cochlea implant, presence of tinnitus and vertigo, medication, noise exposure, pathologic changes in the ear, and other relevant clinical manifestations. Otoscopy, physical examination, pure-tone audiometry (at frequencies from 250 to 8000 Hz), distortion product otoacoustic emission (DPOAE), 40Hz auditory event related potential (40Hz AERP), auditory steady state responses (ASSR) and auditory brainstem responses (ABR) were performed. Immittance testing was applied so that middle-ear functions could be evaluated. Magnetic resonance imaging and/or high-resolution temporal bone computed tomography were performed to evaluate the inner ear structure. Cognitive evaluation was performed using selected subclasses from a Chinese revised version of Griffiths Mental Development Scales (GMDS, 0-8 years of age) prior to cochlea implant operation[1].

***Whole exome and Sanger sequencing***

Blood samples (~3-5 ml) were drawn from 6 participants so that genomic DNA could be extracted with the Genomic DNA isolation kit (QIAGEN). Paternity was confirmed by genotype analysis of 19 informative short tandem repeats (STRs) using Goldeneye ™

20A kit (Peoplespot, Beijing, China) [2], yielding a probability of paternity of 0.999999 (assuming a prior probability of 0.50). Exome capture was performed in pedigrees 1 and 2, including the two probands and their parents, by BGI–Shenzhen using NimbleGen SeqCap EZ Human Exome Library v2.0 (Roche NimbleGen, Inc., Madison, WI, USA) according to the manufacturer's protocols, and sequencing was performed using a HiSeq2000 platform (Illumina, San Diego, CA, USA). Illumina base calling Software 1.7 was used with default parameters to process the raw image files and to sequence the individual products as 90-bp paired-end reads. The sequenced reads were aligned to the human genome reference (UCSC hg19 version, build37.1) using SOAP aligner/SOAP2 [3]. SNP or indels were called using Soapsnp [4] software and bwa [5], respectively. The alignment results were identified using GATK [6] to identify the breakpoints. DNA primers for amplifying *ATP6V1B2* were as follows：

exon-1-F(5'→3'): 5'GGCCGCCTTGGTATATCTGC 3'

exon-1-R(5'→3'): 5'TCTTGCCCTCAGCCCATCTC 3'

exon-2-F(5'→3'): 5'AGCTATTTGAGTGTACCGTATTC 3'

exon-2-R(5'→3'): 5'CAGCTCAGGCAAAAATCTTA 3'

exon-3,4-F(5'→3'): 5'AGCAAAAACCTGTGTCTGTGAA 3'

exon-3,4-R(5'→3'): 5'GCAGCCGGTAACCAATCTCT 3'

exon-5-F(5'→3'): 5'TGCTCATTATATCAAAAGTAGT 3'

exon-5-R(5'→3'): 5'GAGCGTGTTGCCTATCTAC 3'

exon-6-F(5'→3'): 5'TGCTTTATGTAGTTCTGGTCTT 3'

exon-6-R(5'→3'): 5'AAGGGTAAGCAAGAAGACAT 3'

exon-7-F(5’→3’): 5'TGCAAAAAGTACAAAATAGT 3'

exon-7-R(5’→3’): 5'AAAATTCAACTATATATGTACA 3'

exon-8-F(5’→3’): 5'CCGGCTCTTTGTTCTGTTGG 3'

exon-8-R(5’→3’): 5'AATTTGCCCAGTGCCAGATAAC 3'

exon-9-F(5’→3’): 5'TGCTTTACTCTCCTCAATTC 3'

exon-9-R(5’→3’): 5'AGTTTCCTTTCTGATTTATTAA 3'

exon-10-F(5’→3’): 5'TGCCCCATTGTCAACATTTT 3'

exon-10-R(5’→3’): 5'AGGCAAATAACCAAGATAAACTC 3'

exon-11-F(5’→3’): 5'CAGAACATTGGCAGCATCAC 3'

exon-11-R(5’→3’): 5'GCCGACACCAGAATTTAAGAT 3'

exon-12-F(5’→3’): 5'TGCCAGGAAGAGACAGTAGGAT 3'

exon-12-R(5’→3’): 5'AGGCTGCTTTGGTTCTAATGA 3'

exon-13-F(5’→3’): 5'TTCCTTCTATGGGCTCTTGTGA 3'

exon-13-R(5’→3’): 5'AGGAGAAGCCAGAGGGTTAAG 3'

exon-14-F(5’→3’): 5'GCCGCCTCTTCCCTCTTCTC 3'

exon-14-R(5’→3’): 5'TCCATCCCTGCAGCACTTTAA 3'

***Molecular epidemiology analysis of the de novo mutation in ATP6V1B2***

We used the restriction enzyme Taq I (Cat#:R0149L, New England Biolabs, Ipswich, MA,USA) to perform the molecular epidemiology analysis on the *de novo ATP6V1B2* c.1516 C>T mutation in 1053 ethnically-matched normal hearing controls. The primers were as follows:

Forward (5’→3’): 5'AGTTGGAAGTCATTTGCATTTAT3'

Reverse (5’→3’): 5'TGCAAAAGGAATAAAGAAAACA3'

This primer pair used for the restriction enzyme reaction was designed around *ATP6V1B2* c.1516 C, which amplified a 252-bp fragment. The restriction enzyme Taq I recognizes the ‘TC’ at c.1515 and c.1516. After the Taq I reaction, the 252-bp fragment is cut into two fragments of 165 and 87 bp. However, the mutation c.1516 C>T deletes the Taq I restriction endonuclease site. Thus, for the heterozygous c.1516 C>T mutation, three bands (252, 165 and 87 bp) were detected, while in controls lacking the c.1516 C>T mutation, two bands (165 and 87 bp) were detected.

***Quantitative RT-PCR***

To test the mRNA expression of *ATP6V1B2*, a quantitative RT-PCR experiment was performed in Pedigree 3. The total RNA was extracted from the peripheral blood of case 3 and her parents using the QIAamp® RNA Blood Mini (QIAGEN). Complementary DNA (cDNA) was synthesized using RNA, oligo(dT) primer and reverse transcriptase III (TaKaRa, Dalian, China) according to the manufacturer’s instructions. The primers and probes (GeneCore, Shanghai, China) used for TaqMan MGB probe fluorescence real-time quantitative PCR were as follows:

*ATP6V1B2*-F (5’→3’): 5’-GCCTCTTTTTGAACTTGGCTAATG-3’

*ATP6V1B2*-R (5’→3’):5’-AGCCAGGCGAGGAGTGATAA-3’

*GAPDH*-F (5’→3’): 5’-GGGCTGCTTTTAACTCTGGTAAAG-3’

*GAPDH*-R (5’→3’): 5’- CCATGGGTGGAATCATATTGG-3’

*ATP6V1B2* probe (5’→3’): 5’-CCCAACCATTGAGCG-3’

*GAPDH* probe (5’→3’): 5’-CCTCAACTACATGGTTTAC-3’

The probes were labeled with the fluorescent reporter dye FAM (6-carboxyfluorescein) at the 5’ ends and with the fluorescent NFQ-MGB at the 3’ ends. PCR amplifications were carried out on an ABI STEPONE PLUS (Applied Biosystems, USA) with the following protocol: 10 min at 95°C; 40 cycles of 95°C (15 s) followed by 1 min at 60°C. Fluorescence was measured once per cycle at the end of the 60℃ segment. Each sample was tested three times and data were analyzed using the $2^{-\Delta\Delta CT}$ method.

***Atp6v1b2 knockdown in mouse cochlea***

The animal use protocol was approved by the Emory Institutional Animal Care and Use Committee.

To generate a mouse model lacking *Atp6v1b2* function, we designed a splice-blocking morpholino against exon/intron junction 13. Missplicing events at this location were predicted to generate a truncated protein that lacked the last two exons of *Atp6v1b2*. The splice-blocking morpholino (GeneTools, LLC, Philomath, OR, USA) was Morphlino *Atp6v1b2* 5′-CGCATACTGGAGGAGAGAACAGGCC-3′.

***Surgery methods***

Postnatal day 1 or 2 mice were cooled on ice. A skin incision was made behind the ear to expose the cochlea. The tympanic membrane and auditory ossicles were used as landmarks during the surgery. Glass micropipettes (tip size 10–15 μm) were controlled by a micromanipulator to penetrate the lateral wall at the basal turn of the cochlea, close to the stapedial artery, into the scala media. The injection was performed using a Picospritzer III pressure system (Parker Hannifin, NY, USA) for injecting a small volume of fluid (approximately 1.0 μl). The injection was monitored visually by including a dye (fast green; Sigma-Aldrich, St. Louis, MO, USA), which was visible in bright light in the

solution. The skin incision was stitched by a nonabsorbable 8-0 (EP 0.4) suture (S&T AG, Neuhausen, Switzerland). Pups were allowed to recover on a 37°C warming pad.

*Atp6v1b2* specific morpholino oligomer was microinjected (0.05 μg/μl to 5.0 μg/μl) into the scala media of the basal turn of mouse cochlea before postnatal day 3(P3), and we found the lowest dose that induced missplicing events is 0.5 μg. Auditory physiology and molecular biology experiments were carried out on mice injected with 1 μL of 0.5 μg *Atp6v1b2* morpholino. For phenotypic assessment, *Atp6v1b2* morpholino-injected mice were compared to stage-matched mice injected with the same concentration (0.5 μg/μl) of a scrambled control morpholino 5′-AGCTCCAGACGTGAGAGAGAGCGAC-3′ (GeneTools, LLC, Philomath, OR, USA). The *Atp6v1b2*-specific morpholino produced the phenotype of severe sensorineural hearing loss.

The efficacy of *Atp6v1b2* knockdown by the specific morpholino was assessed by RT-PCR analysis of morpholino-induced intron retention. Three days after cochlea injection with morpholinos, the membranous tissues containing membranous labyrinth, spiral ligament, and stria vascularis were taken under the anatomy microscope. Using Ambion RNAqueous® Midi kit (Cat#: AM1912, Life Technologies, Carlsbad, CA, USA), total RNA was extracted from mouse cochlea tissues. To detect RNA quality, the RNA yield and A260:A230 ratios were determined using the NanoDrop® ND-1000 spectrophotometer (NanoDrop Technologies, Wilmington, DE, USA). After removal of DNA by RQ1 RNase-free DNase (Cat#: M6101,Promega, Fitchburg, Wisconsin,USA), single-stranded cDNA templates were synthesized with RNA (500 ng) , an anchored oligo dT primer and Superscript III (Life Technologies). cDNA (1 μl) was then used as template for PCR with primers F: 5′-ATGTGGACAGACAGCTGCAC-3′ and R: 5′-

CTGGGATTCAGGTGTGTCCT-3′. The ß-actin control primers were F: 5′-GAGACCTTCAACACCCCAGC-3′ and R: 5′-CCACAGGATTCCATACCCAA-3′. PCR amplifications were carried out on a GenAmp PCR system 9700 (Applied Biosystems, Beverly, MA, USA) with the following protocol: 2 min denaturation at 94°C; 40 cycles of 94°C (30 s), 60°C (30 s) and 68°C (2 min); followed by 10 min at 72°C. PCR products were visualized by electrophoresis on 1% agarose gels stained with ethidium bromide.

***ABR test***

Mice were anesthetized with ketamine (80 mg/kg) and xylazine (10 mg/kg). Surgical level anesthesia were maintained by a supplementary half-dose of ketamine and xylazine every 60 minutes or as needed depending on the condition of the animals. Rectal temperature was maintained at 37°C with a servo-regulated heating blanket wrapped around animals. The head of the mice was fixed in a head holder with two hollow ear bars. One end of the ear bars was inserted against the external ear canal, and the other end was connected to a high-frequency sound transducer (Intelligent Hearing System, Miami, FL, USA). Three sterile 1/2" 26 gauge needle electrodes (World Precision Instrument, Sarasota, FL) were placed subdermally of each pinna and the vertex respectively to gather signals for the input to a differential amplifier. Typical sound stimuli consisted of click or tone bursts (10 ms in duration and a rise-fall time of 0.5 ms) ranging in frequency from 2.0 to 40.0 kHz. Intensity series (20-100dB SPL) were obtained at 4.0, 8.0, 12.0, 18.0, 24.0, and 32 kHz, and the waveforms recorded were saved for off-line analysis. ABRs were recorded and analyzed by conventional methods using Tucker-Davis System II hardware and software (Tucker-Davis Technologies, Alachua, FL, USA). The hearing threshold was determined by following

the amplitude and latency changes of the wave III or IV in the ABRs until a minimal sound level was determined, at which point just a detectable ABR waveform is observed.

***Flattened whole mount cochlea preparations***

All experiments were performed at room temperature in the cochleae from C57BL/6 wild type mice at postnatal days 2–30 (Charles River Inc., Wilmington, MA, USA) and from *Atp6v1b2*-specific morpholino cochlea knockdown mice. Mice were sacrificed by decapitation, and the temporal bones were rapidly removed and immersed in ice-cold Hanks' balanced salt solution (HBSS; Sigma–Aldrich). The bony capsule of the cochlea was removed with the tip of a 28-gauge needle under a dissection microscope (Stemi DV4, Zeiss, Germany). Starting from the apical turn, we removed the Reissner's membrane from all the cochlea turns using a pair of fine forceps. The lateral wall was kept intact during microdissection. The dissected cochlea turns were cut into ~2 mm segments. The cochlea segments were flattened by placing two micro glass rods at the two ends of each segment. The glass rods were made from broken tips of regular patch-clamp microelectrodes, which were mounted on a drop of Sylgard (Dow Corning Corporation, Elizabethtown, KY, USA) adhered to a glass cover slip. The angle of the mounting allowed the micro glass rods to behave like spring hinges with a downward pressing tendency. Flattened whole-mounted cochlea segments of the cochlea were used in immunolabeling experiments.

***Organotypic cultures***

Cochlea tissues were obtained from C57BL/6 wild-type mice. Only the middle cochlea turn from postnatal day 1 or 2 (P1 or P2) mice was used in the culture to ensure that consistent cochleae were dissected. Tissue dissection procedures were the same as

those used in our published protocol [7]. The explants were placed onto glass coverslips coated with poly-d-lysine (Invitrogen; 500 µg/mL, for 1 h at 20°C) and laminin (Invitrogen; 50 µg/mL in HBSS, for 2 h at 37°C). Cultures were incubated in a defined serum-free culture medium, which contained Dulbecco's modified Eagle's medium (DMED; Sigma–Aldrich), 10 µl/mL N2 supplement (Invitrogen), 25 mM HEPES (Sigma–Aldrich), 6 mg/mL glucose, and 1 mM gentamicin (Sigma–Aldrich), at 37°C with 6.5% $CO_2$ for 72 h. Culture medium was replaced every 24 h. Cultured segments of the cochlea were used in immunolabeling.

***Immunolabeling***

Flattened cochlea samples and cultured segments of the cochlea were fixed in 4% paraformaldehyde in phosphate-buffered solution (PBS) for 20 min. Samples were permeabilized in 0.1% Triton X-100 in PBS for 30 min and blocked in 10% goat serum in PBS for 1 h. Depending on the purpose of the experiment, the following primary antibodies were used: (1) ATP6V1B2 polyclonal antibody (dilution 1:200, 4°C overnight; catalog #15097-1-AP, Proteintech Group Inc. Chicago, IL, USA); (2) MYO6 (dilution 1:200, 4°C overnight, catalog #25-6791, lot #100201, Proteus BioSciences, Ramona, CA, USA); (3) III β-tubulin monoclonal antibody used to label SGNs (dilution 1:500; Cat #MMS-435P, Covance, Princeton, NJ, USA). For flattened cochlea samples, after washing with 0.1% Triton X-100 in PBS three times, samples were incubated with Cy3-conjugated phalloidin (dilution 1:1000; catalog #111-096-003, Jackson Immunoresearch Laboratories, Inc., West Grove, PA, USA) goat anti rabbit secondary antibodies for 1 h at room temperature to visualize immunolabeling results. For cultured segments of the cochlea, after washing with 0.1% Triton X-100 in PBS three times, samples were

incubated with goat anti rabbit secondary antibodies. All samples were mounted in fluoromount-G anti-fading solution (catalog #17984-25, Electron Microscopy Science, PA, USA) and examined with a confocal microscope (Zeiss LSM). Layers of optical sections were superimposed to show a more complete pattern of immunolabeling through multiple layers of cells in the flattened cochlea preparation. For cryosection staining, cochlea tissues were fixed in 4% paraformaldehyde prepared in PBS solution (pH 7.4) overnight at 4°C after they were dissected out carefully using microdissecting tools operated with the aid of a stereomicroscope. Ossified cochleae were decalcified in 120 mM EDTA in PBS for ~3 days, and then dehydrated in 3 M sucrose overnight. The cochlea samples were embedded and stored at –80°C for sectioning. The cochlea sections were cut using a conventional cryosectioning machine (model CM1900; Leica Microsystems, Bannockburn, IL, USA) at a thickness of 8 µm. The same primary and secondary antibodies were used to test the expression patterns of Atp6v1b2, Myo6 and III β-tubulin. Images were acquired using confocal microscopy (Zeiss LSM) and analyzed using Zen 2009 light Edition software by Carl Zeiss.

***Construction of plasmids and transfection into HEK293 cells***

The *ATP6V1B2* cDNA was commercial supplied by (Cat #EX-Z7390-M02, FulenGen,Guangzhou,China). The *ATP6V1B2* gene was amplified by PCR**.** The primers were as follows:

restriction enzymes NheI Forward (5’ → 3’) :

5′ATCATCGCTAGCGCCACCATGGCGCTGCGGGCGATGCGGGG 3′

restriction enzymes SacII Reverse (5’ → 3’) :

5′ATCATCCCGCGGCTAATGCTTTGCAGAGTCTCGAGGG 3′

Oligonucleotide-directed mutagenesis was performed to create constructs of *ATP6V1B2* c.1516C>T mutant plasmids using the TaKaRa MutanBEST Kit (Takara Biotechnology Co., Ltd., Dalian, Liaoning, China), according to the manufacturer's protocol. Primers for *ATP6V1B2* c.1516 C>T mutagenesis were as follows:

Forward (5'→3'): 5′TGAGACTCTGCAAAGCATTAGCCGCGG 3′

Reverse (5'→3'): 5′AGGGTAAAATTCGCTGAGGGTGCTCTGAGG 3′

The wild-type and mutant *ATP6V1B2* cDNA was then cloned into the pIRES2-EGFP expression vector to construct the recombination plasmids. The plasmids were identified by PCR and sequencing.

Four groups of plasmids were transfected into the human embryonic kidney (HEK) 293 cell line: empty vector pIRES2-EGFP; pIRES2-EGFP-*ATP6V1B2*; 1:1 mixture of pIRES2-EGFP-*ATP6V1B2* and pIRES2-EGFP-*ATP6V1B2* p.Arg506X and pIRES2-EGFP-*ATP6V1B2* p.Arg506X. Transfections were carried out with the lipofectamine™ 2000 transfection reagent (Cat #11668019, Invitrogen) according to the manufacturer's instructions. The transfection was verified by western blotting.

***Western blot analyses of ATP6V1B2-encoding V-ATPase obtained from cochleae and transfected cells***

Cochleae without the bony shell and cultured HEK-293 cells were homogenized in RIPA lysis buffer (catalog #20–188, EMD Millipore, Billerica, MA,USA) supplemented with protease inhibitor cocktail (Catalog #539134, CalBiochem, CA,USA). Samples were centrifuged at 16,000 × *g* for 20 min (4°C). Total protein concentrations were measured using the bicinchoninic acid protein assay kit (Pierce, Rockford, IL, USA). Equal amounts of protein (5 μg) were loaded in 4–20% SDS-PAGE gels for electrophoresis.

Proteins were transferred to Hybond ECL nitrocellulose films (Amersham Pharmacia Biotech, Piscataway, NJ, USA). The same antibodies used for immunolabeling were used for Western blotting. Protein loading was verified by detecting a housekeeping protein, ß-actin (Chemicon, Temecula, CA, USA). The dilution factors of antibodies against *ATP6V1B2* and actin were 1:500 and 1:1000, respectively. Results were visualized after processing films with Supersignal West Femto chemiluminescent substrate (Kodak XAR-5 film, Pierce, IL). Atp6v1b2 protein expression levels were quantified on digitized images by normalizing to the band intensity of the ß-actin in corresponding lanes using Image J software package. Details were described previously [8]. Four different batches of *Atp6v1b2* cochlea knockdown mice and 4 batches of HEK 293 cells were included in the analyses.

***ATPase hydrolysis activity***

We used the QuantiChrom ATPase/GTPase Assay Kit (Catalog #DATG-200,BioAssay Systems, Hayward, CA, USA) to detect the ATPase hydrolysis activity [9]. The assay is based on the following reactions: ATPases catalyze the decomposition of ATP into ADP and free phosphate ion; then, liberated phosphate and malachite green reagent form a stable dark green color, which can be measured on a plate reader (600–660 nm). Double distilled water was used as controls. To obtain the enzyme, HEK-293 cells transfected with the indicated constructs (empty vector; *ATP6V1B2*; 1:1 mixture of *ATP6V1B2* and p.Arg506X; and *ATP6V1B2* p.Arg506X) were incubated in buffer A supplemented with protease inhibitor cocktail (Catalog #539134, CalBiochem) and subjected to a freeze-thaw process repeated three times on dry ice. Samples were centrifuged at 16,000 × *g* for 20 min (4°C). Total enzyme concentrations were measured

using the bicinchoninic acid protein assay kit (Pierce). The ATPase hydrolysis activity was measured as directed by the manufacturer. In each run, three wells were assayed for each plasmid, and the experiments were repeated three times independently. Therefore, the data in each group represented a sample size (n) of 3 × 3 = 9 trials for each plasmid and control.

***pH measurements of lysosomes***

An acidotropic fluorescent probe, Lysosensor yellow/blue dextran (10,000 MW, Molecular Probes, Eugene, OR, USA) was used as a pH indicator of lysosomes. HEK293 cells were grown to 80% confluence, trypsinized and harvested ($3 \times 10^6$ cells). The pH calibration curve was generated as described previously [10]. Briefly, normal HEK293 cells were labeled with Lysosensor dextran (1 mg/mL) and incubated for 2 h at 37°C. Excess dye was removed with cold PBS washes. The cells were then treated with MES calibration buffer solutions (5 mM NaCl, 115 mM KCl, 1.2 mM MgSO4 and 25 mM MES, pH ranging from 3.5 to 7.0) containing 10 μM monensin and 10 μM nigericin. The emission scan was measured using excitation at 360 nm, with both emission and excitation bandwidths set to 4 nm. Subsequently, the fluorescence emission intensity ratios at 460 nm and 528 nm, respectively, were calculated. The measured data points (intensity ratio at 460 nm/528 nm) were fitted to a Boltzmann equation (ORIGIN, Microcal software Inc, Northampton, MA, USA). Measurements of lysosomal pH of HEK293 cells transfected with indicated constructs (empty vector; *ATP6V1B2*; 1:1 mixture of *ATP6V1B2* and p.Arg506X; and *ATP6V1B2* p.Arg506X) were performed as described above, with the exception that the cells were resuspended in MES buffer (pH 7.76). The emission intensity ratio at 460 nm and 528 nm were converted to an absolute

value of lysosomal pH by comparison with the calibration curve generated above.

## Web Resources

The URLs for data presented herein are as follows:

SIFT：http://sift.jcvi.org/

PolyPhen：http://genetics.bwh.harvard.edu/pph2/index.shtml

Mutationtaster：http://www.mutationtaster.org/

GenBank, http://www.ncbi.nlm.nih.gov/Genbank/

HereditaryHearing Loss homepage,http://hereditaryhearingloss.org

HUGOGeneNomenclatureCommittee,http://www.genenames.org

Morton Cochlear EST Database,

http://www.brighamandwomens.org/bwh_hearing/human-cochlear-ests.aspx

Online Mendelian Inheritance in Man (OMIM), http://www.ncbi.nlm.nih.gov/Omim/

UCSC Genome Browser, http://genome.ucsc.edu/cgi-bin/hgGateway

Image J software package, http://rsbweb.nih.gov/ij/features.html

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