Supplementary Online Content

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This supplementary material has been provided by the authors to give readers additional information about their work.
**eTable 1. Risk Factors of Hearing Loss in 238 Patients With Failed Hearing Screening**

| Risk factors                                 | No. (%)   |
|----------------------------------------------|-----------|
| NICU stay ≥ 5 days                           | 210 (88.2)|
| Oxygen exposure                              | 71 (29.8) |
| Maternal complication during pregnancy       | 69 (29.0) |
| Low birth weight                             | 67 (28.2) |
| Sepsis                                       | 62 (26.1) |
| Perinatal asphyxia                           | 50 (21.0) |
| Mechanical ventilation                       | 45 (18.9) |
| Craniofacial malformation                    | 29 (12.2) |
| Severe hyperbilirubinemia                    | 20 (8.4)  |
| Cytomegalovirus infection                    | 14 (6.4)  |
| Bacterial meningitis                         | 10 (4.2)  |
| Family history of hearing loss               | 4 (1.7)   |
| Consanguineous parents                       | 1 (0.4)   |

*a Multiple risk factors in one patient were counted separately.  b A total of 220 of 238 patients conducted tests for cytomegalovirus.*
# eTable 2. Genes With Pathogenic/Likely Pathogenic Variants Identified in 8078 Patients

| Gene   | Disease [MIM number]                                                                 | Inheritance | No. (%)<sup>a</sup> |
|--------|-------------------------------------------------------------------------------------|-------------|---------------------|
| GJB2   | Deafness, autosomal recessive 1A [220290]                                            | AR/DD       | 58 (64.4)           |
| SLC26A4| Deafness, autosomal recessive 4, with enlarged vestibular aqueduct [600791], Pendred syndrome [274600] | AR          | 17 (18.9)           |
| COL11A1| Deafness, autosomal dominant 37 [618533], Marshall syndrome [154780], Stickler syndrome, type II [604841] | AD          | 2 (2.2)             |
| COL2A1 | Knies dysplasia [156550], SED congenita [183900], Spondyloperipheral dysplasia [271700], Stickler syndrome, type I [108300] | AD          | 2 (2.2)             |
| CHD7   | CHARGE syndrome [214800], Hypogonadotropic hypogonadism 5 with or without anosmia [612370] | AD          | 1 (1.1)             |
| CREBBP | Menke-Hennekam syndrome 1 [618332], Rubinstein-Taybi syndrome 1 [180849]            | AD          | 1 (1.1)             |
| FLNA   | Frontometaphyseal dysplasia 1 [305620], Otopalatodigital syndrome, type I [311300], Otopalatodigital syndrome, type II [304120] | XLD/XLR     | 1 (1.1)             |
| KCNQ4  | Deafness, autosomal dominant 2A [600101]                                            | AD          | 1 (1.1)             |
| MAF    | Ayme-Gripp syndrome [601088]                                                        | AD          | 1 (1.1)             |
| MYH9   | Deafness, autosomal dominant 17 [603622], Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss [155100] | AD          | 1 (1.1)             |
| OTOGL  | Deafness, autosomal recessive 84B [614944]                                          | AR          | 1 (1.1)             |
| PEX1   | Heimler syndrome 1 [234580], Peroxisome biogenesis disorder 1A (Zellweger) [214100], Peroxisome biogenesis disorder 1B (NALD/IRD) [601539] | AR          | 1 (1.1)             |
| PTPN11 | LEOPARD syndrome 1 [151100], Noonan syndrome 1 [163950]                            | AD          | 1 (1.1)             |
| RUNX2  | Cleidocranial dysplasia [119600]                                                    | AD          | 1 (1.1)             |
| TRRAP  | Developmental delay with or without dysmorphic facies and autism [618454]           | AD          | 1 (1.1)             |
| **Total** | NA                                                                                 | NA          | 90 (100)            |

Abbreviations: AD, autosomal dominant inheritance; AR, autosomal recessive inheritance; DD, digenic dominant
inheritance; NA, not applicable; XLD, X-linked dominant inheritance; XLR, X-linked recessive inheritance. *
Percentage was the proportion of the gene in all genes.
**eTable 3. Results of Genetic Tests and Relationship With Hearing Screening in 8078 Patients**

| Gene       | Variant(s) | Zygosity | Hearing screening, No. (%) | Total |
|------------|------------|----------|---------------------------|-------|
|            |            |          | Positive | Negative |       |
| **GJB2**   |            |          |          |          |       |
| c.109G>A   | Hom        | 23 (68)  | 11 (32)  | 34      |
| c.109G>A/c.176_191del | Het/Het | 1 (100)  | 0 (0)    | 1       |
| c.109G>A/c.235del | Het/Het | 5 (63)   | 3 (38)   | 8       |
| c.109G>A/c.299_300del | Het/Het | 1 (100)  | 0 (0)    | 1       |
| c.139G>T/c.235del | Het/Het | 1 (100)  | 0 (0)    | 1       |
| c.176_191del/c.235del | Het/Het | 1 (100)  | 0 (0)    | 1       |
| c.235del   | Hom        | 8 (80)   | 2 (20)   | 10      |
| c.235del/c.299_300del | Het/Het | 1 (100)  | 0 (0)    | 1       |
| **SLC26A4** |            |          |          |          |       |
| c.589G>A/c.1174A>T | Het/Het | 1 (50)   | 1 (50)   | 2       |
| c.600+2T>A/c.919-2A>G | Het/Het | 1 (100)  | 0 (0)    | 1       |
| c.919-2A>G | Hom        | 2 (50)   | 2 (50)   | 4       |
| c.919-2A>G/c.1343C>T | Het/Het | 1 (100)  | 0 (0)    | 1       |
| c.919-2A>G/c.1707+5G>A | Het/Het | 1 (100)  | 0 (0)    | 1       |
| c.919-2A>G/c.1975G>C | Het/Het | 0 (0)    | 1 (100)  | 1       |
| c.919-2A>G/c.2168A>G | Het/Het | 1 (50)   | 1 (50)   | 2       |
| c.1174A>T/c.1229C>T | Het/Het | 1 (100)  | 0 (0)    | 1       |
| c.1226G>A   | Hom        | 0 (0)    | 1 (100)  | 1       |
| c.1229C>T/c.1975G>C | Het/Het | 1 (100)  | 0 (0)    | 1       |
| c.1229C>T/c.2168A>G | Het/Het | 0 (0)    | 1 (100)  | 1       |
| c.2168A>G   | Hom        | 0 (0)    | 1 (100)  | 1       |
| **COL2A1** | c.2050-1G>C | Het     | 1 (100)  | 0 (0)    | 1       |
| c.2818C>T   | Het        | 1 (100)  | 0 (0)    | 1       |
| **COL11A1** | c.1798C>T  | Het     | 1 (100)  | 0 (0)    | 1       |
| c.3816+2dup | Het        | 0 (0)    | 1 (100)  | 1       |
| **CHD7**   | c.6165_6166del | Het | 1 (100)  | 0 (0)    | 1       |
| **CREBBP** | c.4660A>T  | Het     | 1 (100)  | 0 (0)    | 1       |
| **FLNA**   | c.620C>T   | Het     | 1 (100)  | 0 (0)    | 1       |
| **KCNQ4**  | c.2039C>T  | Het     | 0 (0)    | 1 (100)  | 1       |
| **MAF**    | c.161C>T   | Het     | 0 (0)    | 1 (100)  | 1       |
| **MYH9**   | c.534dup   | Het     | 1 (100)  | 0 (0)    | 1       |
| **OTOGl**  | c.2833C>T/c.2911del | Het/Het | 1 (100)  | 0 (0)    | 1       |
| **PEX1**   | c.2050C>T/c.3043G>T | Het/Het | 1 (100)  | 0 (0)    | 1       |
| **PTPN11** | c.1510A>G  | Het     | 1 (100)  | 0 (0)    | 1       |
| **RUNX2**  | c.860-1G>C | Het     | 1 (100)  | 0 (0)    | 1       |
| **TRRAP**  | c.3316G>A  | Het     | 1 (100)  | 0 (0)    | 1       |
| **Total**  |            | /       | 63 (70)  | 27 (30)  | 90     |

Abbreviations: Het, heterozygous variants; Hom, homozygous variants.

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**eTable 4.** Characteristics of 25 Patients Lost to Follow-up Compared With 240 Patients Who Completed Follow-up

| Characteristics          | Lost to follow-up, No. (%) (n=25) | Completed follow-up, No. (%) (n=240) | P value |
|-------------------------|-----------------------------------|--------------------------------------|---------|
| Sex                     |                                   |                                      |         |
| Male                    | 14 (56.0)                         | 153 (63.8)                           | .45     |
| Female                  | 11 (44.0)                         | 87 (36.2)                            |         |
| Gestational age         |                                   |                                      |         |
| < 28 weeks              | 1 (4.0)                           | 7 (2.9)                              | .26     |
| 28-31 weeks             | 3 (12.0)                          | 18 (7.5)                             |         |
| 32-36 weeks             | 8 (32.0)                          | 44 (18.3)                            |         |
| ≥ 37 weeks              | 13 (52.0)                         | 171 (71.3)                           |         |
| Birth weight            |                                   |                                      |         |
| < 1000 g                | 1 (4.0)                           | 6 (2.5)                              | .006    |
| 1000-1499 g             | 2 (8.0)                           | 16 (6.7)                             |         |
| 1500-2499 g             | 11 (44.0)                         | 36 (15.0)                            |         |
| 2500-3999 g             | 11 (44.0)                         | 171 (71.2)                           |         |
| ≥ 4000 g                | 0 (0.0)                           | 11 (4.6)                             |         |
| Cesarean                | 9 (36.0)                          | 104 (43.3)                           | .48     |
| Failed NBHS             | 24 (96.0)                         | 214 (89.2)                           | .49     |
| Positive genetic findings* | 5 (20.0)                         | 85 (35.4)                            | .12     |

Abbreviations: NBHS, Newborn hearing screening programs. * Positive genetic findings were patients identified with hearing loss related genes.
### eTable 5. Results of Follow-ups in Patients Who Failed Hearing Screening and Had Genetic Findings

| Gene    | Variant(s)                  | Zygosity | AI, M (range), (months) | AD, M (range), (months) | HL, No.* | Treatment, No. | Non-HL, No. | Total, No. |
|---------|------------------------------|----------|------------------------|-------------------------|----------|----------------|-------------|------------|
|         |                              |          |                        |                         | M/M      | S/P            | CI          |            |
|         |                              |          |                        |                         |          |                | HA/ST       |            |
|         |                              |          |                        |                         |          |                | None        |            |
|         |                              |          |                        |                         |          |                |             |            |
| **GJB2**| c.109G>A                     | Hom      | 41 (9–64)              | 3 (3–6)                 | 5        | 1              | 6           | 0          | 2          | 4          | 15         | 21         |
|         | c.109G>A/ c.176_191del        | Het/Het  | 20                     | NA                      | NA       | NA             | NA          | NA         | NA         | NA         | 1          | 1          |
|         | c.109G>A/c.235del             | Het/Het  | 21.5 (10–26)           | NA                      | NA       | NA             | NA          | NA         | NA         | NA         | 4          | 4          |
|         | c.109G>A/ c.299_300del        | Het/Het  | 25                     | NA                      | NA       | NA             | NA          | NA         | NA         | NA         | 1          | 1          |
|         | c.139G>T/c.235del             | Het/Het  | 22                     | 4                       | 1        | 0              | 1           | 0          | 0          | 1          | 0          | 1          |
|         | c.176_191del/ c.235del        | Het/Het  | 44                     | 3                       | 0        | 1              | 1           | 0          | 1          | 0          | 1          | 1          |
|         | c.235del                     | Hom      | 26.5 (15–62)           | 3 (1.5–4)               | 2        | 6              | 8           | 4          | 3          | 2          | 0          | 8          |
|         | c.235del/ c.299_300del        | Het/Het  | 25                     | 3                       | 0        | 1              | 1           | 1          | 1          | 0          | 0          | 1          |
|         | c.299_300del                 | Hom      | 53                     | 24                      | 1        | 0              | 1           | 0          | 0          | 1          | 0          | 1          |
| **SLC26A4** | c.589G>A/c.1174A>T          | Het/Het  | 34                     | 4.5                     | 0        | 1              | 1           | 1          | 1          | 0          | 0          | 1          |
|         | c.600+2T>A/c.919-2A>G        | Het/Het  | 64                     | 18                      | 0        | 1              | 1           | 0          | 1          | 0          | 0          | 1          |
|         | c.919-2A>G                   | Hom      | 49.5 (39–60)           | 8 (3–13)                | 0        | 2              | 2           | 2          | 1          | 0          | 0          | 2          |
|         | c.919-2A>G/c.1343C>T         | Het/Het  | 27                     | 3                       | 1        | 0              | 1           | 0          | 1          | 0          | 0          | 1          |
|         | c.919-2A>G/c.1707+5G>A       | Het/Het  | 57                     | 3                       | 0        | 1              | 1           | 1          | 1          | 0          | 0          | 1          |
|         | c.919-2A>G/c.2168A>G         | Het/Het  | 28                     | 3                       | 0        | 1              | 1           | 1          | 1          | 0          | 0          | 1          |
|         | c.1174A>T/c.1229C>T          | Het/Het  | 41                     | 4                       | 0        | 1              | 1           | 0          | 1          | 0          | 0          | 1          |
|         | c.1229C>T/c.1975G>C          | Het/Het  | 47                     | NA                      | NA       | NA             | NA          | NA         | NA         | NA         | 1          | 1          |
| **COL2A1** | c.2050-1G>C                  | Het      | 47                     | NA                      | NA       | NA             | NA          | NA         | NA         | NA         | 1          | 1          |
|         | c.2818C>T                    | Het      | 28                     | 1.5                     | 1        | 0              | 1           | 0          | 0          | 1          | 0          | 1          |
| **COL11A1** | c.1798C>T                  | Het      | 29                     | NA                      | NA       | NA             | NA          | NA         | NA         | NA         | 1          | 1          |
| **CREBBP** | c.4660A>T                  | Het      | 55                     | NA                      | NA       | NA             | NA          | NA         | NA         | NA         | 1          | 1          |
| **FLNA** | c.620C>T                     | Het      | 29                     | NA                      | NA       | NA             | NA          | NA         | NA         | NA         | 1          | 1          |

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| Gene  | Variant(s)          | Zygosity | AI, M (range), (months) | AD, M (range), (months) | HL, No. | Treatment, No. | Non-HL, No. | Total, No. |
|-------|---------------------|----------|-------------------------|-------------------------|---------|----------------|-------------|------------|
|       |                     |          |                         |                         | M/M     | S/P   | Total | Cl | HA/ST | None |         |          |
| MYH9  | c.534dup            | Het      | 45                       | 2.5                     | 0       | 1     | 1     | 1  | 1     | 0    | 0       | 1       |
| OTOGL | c.2833C>T/c.2911del  | Het/Het  | 23                       | 5                       | 1       | 0     | 1     | 0  | 0     | 1    | 0       | 1       |
| PEX1  | c.2050C>T/c.3043G>T  | Het/Het  | 46                       | 5                       | 0       | 1     | 1     | 0  | 0     | 1    | 0       | 1       |
| PTPN11| c.1510A>G           | Het      | 44                       | NA                      | NA      | NA    | NA    | NA| NA    | NA   | 1       | 1       |
| RUNX2 | c.860-1G>C          | Het      | 23                       | 3                       | 1       | 0     | 1     | 0  | 0     | 1    | 0       | 1       |
| TRRAP | c.3316G>A           | Het      | 13                       | 3                       | 1       | 0     | 1     | 0  | 0     | 1    | 0       | 1       |
| Total | NA                  | NA       | 33 (9–64)                | 3 (1.5–24)             | 14      | 18    | 32    | 11| 15    | 13   | 27      | 59      |

Abbreviations: AD, age at diagnosis; AI, age at interview; CI, Cochlear implant; HA, Hearing aids; Het, heterozygosity; HL, hearing loss; Hom, homozygosity; M/M, mild or moderate degree of hearing loss; M, median; NA, not applicable; S/P, severe or profound degree of hearing loss; ST, Speech therapy. * The different degree of hearing in both ears in one patient was counted according to the more severe one.
**eTable 6. Hearing Loss Diagnosed Without Genetic Findings**

| No.  | Sex | GA (weeks) | AI (months) | AD (months) | Main clinical diagnosis                                                                 | Risk factors of hearing loss                                                                 | Degree       | Laterality | Treatment                        |
|------|-----|------------|-------------|-------------|---------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------|--------------|------------|----------------------------------|
| P-19 | M   | 40<sup>+</sup> | 56          | 30          | Cholestasis, cleft palate                                                              | NICU stay ≥ 5 d, perinatal asphyxia                                                         | Moderate     | Bilateral | Hearing aids/ speech therapy     |
| P-95 | M   | 39<sup>+</sup> | 38          | 3           | Hyperbilirubinemia, congenital heart disease                                         | Severe hyperbilirubinemia                                                                    | Moderate/ severe | Bilateral | Hearing aids                      |
| P-118| F   | 37<sup>+</sup> | 33          | 6           | Congenital malformation (fish lip), intracranial hemorrhage, congenital heart disease | NICU stay ≥ 5 d, maternal complication during pregnancy, low birth weight, craniofacial malformation | Mild         | Bilateral | None                             |
| P-233| M   | 35<sup>+</sup> | 11          | 3           | Hyperbilirubinemia, preterm, congenital heart disease                               | NICU stay ≥ 5 d                                                                            | Mild         | Unilateral | None                             |
| P-28 | M   | 39<sup>+</sup> | 53          | 3           | Sepsis, lebanese fossa fistula                                                       | NICU stay ≥ 5 d, oxygen exposure, sepsis, mechanical ventilation                            | Mild         | Bilateral | None                             |
| P-259| M   | 41<sup>+</sup> | 10          | 3           | Neonatal hypoglycemia, sepsis                                                       | NICU stay ≥ 5 d, sepsis, maternal complication during pregnancy                             | Mild         | Unilateral | None                             |
| P-194| M   | 38<sup>+</sup> | 27          | 3           | Absence of eyeball (left eye), sepsis, craniofacial malformation                  | NICU stay ≥ 5 d, sepsis, craniofacial malformation, cytomegalovirus infections             | Mild         | Unilateral | None                             |
| No.  | Sex | GA (weeks) | AI (months) | AD (months) | Main clinical diagnosis                                      | Risk factors of hearing loss                                                                 | Degree    | Laterality | Treatment |
|------|-----|------------|-------------|-------------|----------------------------------------------------------------|------------------------------------------------------------------------------------------|-----------|------------|-----------|
| P-243| F   | 40<sup>+</sup> | 9           | 3           | Seizure, congenital heart disease                                | NICU stay ≥ 5 d, oxygen exposure                                                          | Moderate  | Bilateral  | None      |
| P-146| M   | 21<sup>+</sup> | 31          | 3           | Preterm, neonatal encephalopathy                                 | NICU stay ≥ 5 d, oxygen exposure, maternal complication during pregnancy, low birth weight, sepsis, perinatal asphyxia, mechanical ventilation, craniofacial malformation, cytomegalovirus infections | Mild      | Bilateral  | None      |
| P-162| F   | 38<sup>+</sup> | 17          | 3           | Small for gestational age, congenital dysplasia joint, subdural hemorrhage | NICU stay ≥ 5 d, maternal complication during pregnancy, low birth weight, craniofacial malformation | Moderate  | Unilateral | None      |
| P-169| F   | 40<sup>+</sup> | 7           | 7           | Pierre Robin Syndrome                                            | NICU stay ≥ 5 d, oxygen exposure, maternal complication during pregnancy, low birth weight, perinatal asphyxia, mechanical ventilation, craniofacial malformation | Mild/moderate | Bilateral  | Non       |
| P-197| M   | 39<sup>+</sup> | 12          | 8           | Hyperbilirubinemia, subdural hemorrhage                          | Maternal complication during pregnancy, craniofacial malformation                          | Moderate/severe | Bilateral  | None<sup>a</sup> |

<sup>a</sup> Indicates additional treatment

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| No.   | Sex | GA (weeks) | AI (months) | AD (months) | Main clinical diagnosis                          | Risk factors of hearing loss                                                                 | Degree | Laterality | Treatment |
|-------|-----|------------|-------------|-------------|-------------------------------------------------|---------------------------------------------------------------------------------------------|--------|------------|-----------|
| P-235 | M   | 38<sup>1</sup> | 12          | 3           | Hyperbilirubinemia, intraventricular hemorrhage | NICU stay ≥ 5 d, maternal complication during pregnancy, craniofacial malformation          | Mild   | Bilateral  | None      |

Abbreviations: AD, age at diagnosis; AI, age at interview; F, female; GA, gestational age; M, male; * P-197 was suggested for unilateral cochlear implant after auricle reconstruction.