

## Supplemental Appendix

### Title: Treatment and long-term outcome in primary distal Renal Tubular Acidosis.

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## Supplemental Tables

Supplemental table 1: Data points collected

| Supplemental Table 1. Data points collected |   |
|---|---|
| <b>Renal Unit details</b>                   | <b>Laboratory</b>                                 |
| Treating physician                          | Serum creatinine ( $\mu\text{mol/L}$ )            |
| Email address                               | Serum bicarbonate ( $\text{mmol/L}$ )             |
| Centre – City                               | Urinary calcium/creatinine ( $\text{mmol/mmol}$ ) |
| Country                                     | <b>Treatment (mmols/day)</b>                      |
| ERK-Net centre (y/n)                        | Bicarbonate (sodium +/- potassium)                |
| <b>Demographic data</b>                     | Citrate (sodium +/- potassium)                    |
| Patient ID                                  | Others  |
| Gender (F/M)                                | <b>Comorbidities</b>                              |
| Age at presentation (years)                 | Nephrocalcinosis (y/n)                            |
| <b>Genetic information</b>                  | Age at diagnosis (years)                          |
| Gene  | Urolithiasis (y/n)                                |
| Mutation details                            | Age lithiasis was first diagnosed (years)         |
| <b>Auxiometry</b>                           | Hearing loss (y/n)                                |
| Current age (years)                         | Hearing aids prescribed (y/n)                     |
| Current height (cm)                         | Age at prescription (years)                       |
| Current weight (kg)                         | Cochlear implantation (y/n)                       |
|   | Age when implanted (years)                        |

Shown are the data points that were requested in the online form.

Physicians were invited to participate via the following email:

Dear colleagues,

We, the working groups for inherited kidney diseases of ERA-EDTA and ESPN and ERKNet, are aiming at better understanding the long-term outcome of patients with dRTA, as well as current treatment practices. To this end, we would be grateful if you could fill in our brief online survey for each of your patients with a presumed or proven inherited form of dRTA.

To enter the survey, please copy the following Web link to your browser:

<https://www.surveymonkey.de/r/drta>

With the patient file at hand, it should not take more than 5 minutes per patient to complete the survey.

If more than one colleagues from a center respond to this survey, please make sure to enter each patient only once.

The survey should work equally well from a PC, iPad and Smartphone.

We aim to publish the results and physicians contributing to this survey will be sharing into the authorship; either as named members of the "European dRTA consortium" or, if you contribute three or more patients, as named co-authors.

We greatly appreciate your help!

Detlef Bockenhauer and Franz Schaefer

Supplemental table 2: Valid and missing data

| Supplemental Table 2. Variables Valid and Missing data | N     |         |
|--|-------|---------|
|  | Valid | Missing |
| Gender   | 340   | 0       |
| Age at presentation                                    | 336   | 4       |
| Current age  | 340   | 0       |
| Mutation   | 340   | 0       |
| Current weight   | 334   | 6       |
| Height SDS   | 323   | 17      |
| Alkali daily dose                                      | 334   | 6       |
| Serum Creatinine ( $\mu\text{mol/L}$ )                 | 340   | 0       |
| Serum bicarbonate                                      | 340   | 0       |
| Treatment option                                       | 340   | 0       |
| Hypercalciuria   | 312   | 28      |
| Nephrocalcinosis                                       | 339   | 1       |
| Age at diagnosis of Nephrocalcinosis (N=299)           | 261   | 38      |
| Nephrolithiasis  | 331   | 9       |
| Age at diagnosis of Nephrolithiasis (N=79)             | 64    | 15      |
| Hearing loss   | 326   | 14      |
| Hearing aids   | 90    | 0       |
| Age when aids were prescribed                          | 76    | 14      |
| Cochlear Implants                                      | 24    | 2       |
| Age at cochlear implantation                           | 22    | 2       |

Shown are the numbers of patients for whom the requested data were available (“valid”) or not (“missing”)

Supplemental Table 3. Gross Domestic Product (GDP) per capita in the participating countries.

Participating countries were classified in three different groups according to their per capita GDP in 2016 as “Low” (<10.000 USD/year), “Medium” (10.000-35.000 USD/year) or “High” (>35.000 USD/year).

| <b>Supplemental Table 3. Gross Domestic Product (GDP) per capita in the year 2016</b> |                               |                       |
|---|-------------------------------|-----------------------|
| Country Name  | GDP per capita (current US\$) | GPD (low/medium/high) |
| India   | 1709.59                       | LOW                   |
| Egypt, Arab Rep.  | 3477.85                       | LOW                   |
| Kosovo  | 3661.43                       | LOW                   |
| Belarus   | 4989.43                       | LOW                   |
| Iran, Islamic Rep.  | 5219.11                       | LOW                   |
| Macedonia, FYR  | 5237.15                       | LOW                   |
| South Africa  | 5274.55                       | LOW                   |
| Serbia  | 5426.20                       | LOW                   |
| Montenegro  | 7028.93                       | LOW                   |
| Russian Federation  | 8748.37                       | LOW                   |
| Turkey  | 10862.60                      | MEDIUM                |
| Croatia   | 12149.19                      | MEDIUM                |
| Poland  | 12414.10                      | MEDIUM                |
| Lithuania   | 14912.69                      | MEDIUM                |
| Greece  | 17890.57                      | MEDIUM                |
| Portugal  | 19871.72                      | MEDIUM                |
| Saudi Arabia  | 20028.65                      | MEDIUM                |
| Spain   | 26616.49                      | MEDIUM                |
| Italy   | 30668.98                      | MEDIUM                |
| France  | 36857.12                      | HIGH                  |
| Israel  | 37180.53                      | HIGH                  |
| United Kingdom  | 40412.03                      | HIGH                  |
| Belgium   | 41271.48                      | HIGH                  |
| Germany   | 42161.32                      | HIGH                  |
| Netherlands   | 45637.89                      | HIGH                  |
| Australia   | 49755.32                      | HIGH                  |
| Sweden  | 51844.76                      | HIGH                  |
| Denmark   | 53578.76                      | HIGH                  |
| Switzerland   | 79887.52                      | HIGH                  |



Supplemental Table 4. Participating countries.

**Supplemental table 4: response rate according to country of the submitting clinician**

Shown is the number of patients according to submitting country

| Table 1. Participating countries. |                   |              |                   |
|-----------------------------------|-------------------|--------------|-------------------|
| Country                           | Patients Included | Country      | Patients Included |
| Italy                             | 59                | Greece       | 6                 |
| France                            | 39                | Lithuania    | 5                 |
| Germany                           | 36                | Portugal     | 5                 |
| UK                                | 33                | Denmark      | 4                 |
| Turkey                            | 23                | Saudi Arabia | 4                 |
| India                             | 22                | Sweden       | 4                 |
| Poland                            | 12                | Belarus      | 3                 |
| Spain                             | 12                | Croatia      | 3                 |
| Netherlands                       | 10                | Switzerland  | 2                 |
| Iran                              | 9                 | Israel       | 1                 |
| Australia                         | 8                 | Kosovo       | 1                 |
| Egypt                             | 7                 | Montenegro   | 1                 |
| Macedonia                         | 7                 | Serbia       | 1                 |
| Russia                            | 7                 | South Africa | 1                 |
| Belgium                           | 6                 |              |                   |

## Supplemental Table 5. Data summary of the whole cohort

Genetic results included when available. N.M.I.: No pathogenic mutation identified, AR: Autosomal Recessive, AD: Autosomal Dominant, H: homozygous, h: heterozygous, ch: compound heterozygous Mutation details as reported by the submitting clinicians. No formal genetic report was submitted and some results may have been obtained on a research basis only, without confirmation by a clinical genetic laboratory. Thus, genetic data need to be interpreted with caution. In some patients, clinicians had only noted the underlying gene, but were unable to retrieve the exact mutation details (“exact genotype not retrievable”). “Uca/Ucr normalised” refers to ratio of the concentrations of calcium and creatinine in the urine, normalised to the upper limit of normal of the age-appropriate reference range. Thus, a value >1.0 reflects hypercalciuria. The columns “Hearing aids (years)” and “cochlear implants (years)” detail the age at which this was prescribed or performed.

| Gender | Age<br>Diagnosis<br>(year) | Gen      | Inheritance | Trait | c.DNA                                     | Protein result                            | Weight<br>(kg) | Height<br>(cm) | Height<br>SDS | Creatinine (mEq/<br>μmol/L) | Alkali<br>serum dose<br>(mEq/kg/day) | Age at<br>last<br>Follow<br>Up | eGFR<br>(ml/<br>min) | CKD<br>Stages<br>I-V | HCO <sub>3</sub> <sup>-</sup><br>(mmol/L) | Treatment | Hyper-<br>calciuria | Metabolic<br>Control | Nephro-<br>calcinosis | Nephro-<br>lithiasis | Hearing<br>loss | Hearing<br>aids | Hearing<br>aids<br>prescription<br>(age) | Cochlear<br>Implantation | Cochlear<br>prescription<br>(years) | Time<br>of FU<br>(years) |     |
|--------|----------------------------|----------|-------------|-------|---|---|----------------|----------------|---------------|-----------------------------|--------------------------------------|--------------------------------|----------------------|----------------------|---|-----------|---------------------|----------------------|-----------------------|----------------------|-----------------|-----------------|--|--------------------------|-------------------------------------|--------------------------|-----|
|        |                            |          |             |       |   |   |                |                |               |                             |                                      |                                |                      |                      |   |           |                     |                      |                       |                      |                 |                 |  |                          |                                     |                          |     |
| Female | 0.2                        | ATP6V0A4 | AR          | H     | c.[823A>C]+<br>c.[823A>C]                 | p.[Thr275Pro]+<br>p.[Thr275Pro]           | 5.6            | 61.5           | 1.58          | 17.7                        | 2.1                                  | 0.2                            | 127                  | CKD-1                | 22.5                                      | Both      | No                  | Yes                  | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 0.0                      |     |
| Male   | 0.2                        | ATP6V0A4 | AR          | H     | c.[2257C>T]+<br>c.[2257C>T]               | p.[Gln753*]+<br>p.[Gln753*]               | 7.7            | 71             | -0.98         | 17.7                        | 3.9                                  | 0.9                            | 146                  | CKD-1                | 22.6                                      | Both      | No                  | Yes                  | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 0.7                      |     |
| Female | 0.16                       | ATP6V0A4 | AR          | H     | c.[639+1G>A]+<br>c.[639+1G>A]             | p[?]+p[?]                                 | 7.7            | 70.5           | -3.2          | 24                          | 2.5                                  | 1.5                            | 107                  | CKD-1                | 20  | HCO3      | No                  | No                   | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 1.3                      |     |
| Male   | 0.2                        | ATP6V0A4 | AR          | H     | c.[16C>T]+<br>c.[16C>T]                   | p.[Arg6*]+<br>p.[Arg6*]                   | 10             | 83             | -1.32         | 26                          | 3.4                                  | 2                              | 117                  | CKD-1                | 20  | Citrate   | No                  | No                   | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 1.8                      |     |
| Female | 0.9                        | ATP6V0A4 | AR          | H     | c.[2035G>T]+<br>c.[2035G>T]               | p.[Asp679Tyr]+<br>p.[Asp679Tyr]           | 11.3           | 84             | -0.62         | 49                          | 2.7                                  | 2                              | 63                   | CKD-2                | 27.2                                      | HCO3      | Yes                 | No                   | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 1.1                      |     |
| Male   | 0.1                        | ATP6V0A4 | AR          | H     | c.[2257+1G>A]+<br>c.[2257+1G>A]           | p.[?]+p.[?]                               | 13.6           | 91.3           | 0.94          | 34                          | 4.7                                  | 2                              | 98                   | CKD-1                | 20  | Citrate   | Yes                 | No                   | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 1.9                      |     |
| Male   | 0.25                       | ATP6V0A4 | AR          | H     | c.[2257C>T]+<br>c.[2257C>T]               | p.[Gln753*]+<br>p.[Gln753*]               | 9.6            | 83             | -1.32         | 31                          | 5                                    | 2                              | 98                   | CKD-1                | 18.7                                      | Both      | No                  | No                   | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 1.8                      |     |
| Female | 0                          | ATP6V0A4 | AR          | CH    | c.[2308C>T]+<br>c.[2257C>T]               | p.[Arg770*]+<br>p.[Gln753*]               | 14             | 93.7           | 2.22          | 29                          | 5.4                                  | 2                              | 118                  | CKD-1                | 24  | Citrate   | No                  | Yes                  | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 2.0                      |     |
| Female | 0.08                       | ATP6V0A4 | AR          | H     | c.[639+1G>A]+<br>c.[639+1G>A]             | p[?]+p[?]                                 | 11.5           | 87             | 0.23          | 24                          | 5.9                                  | 2                              | 132                  | CKD-1                | 22  | Both      | No                  | Yes                  | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 1.9                      |     |
| Male   | 1.1                        | ATP6V0A4 | AR          | H     | c.[596T>A]+<br>c.[596T>A]                 | p.[Leu199*]+<br>p.[Leu199*]               | 11.5           | 74             | -4.12         | 40                          | 9.1                                  | 2.2                            | 67.5                 | CKD-2                | 22  | Citrate   | Yes                 | No                   | Yes                   | Yes                  | No              | No              | No                                       | No                       | No                                  | 1.1                      |     |
| Female | 0.1                        | ATP6V0A4 | AR          | H     | c.[1107delC]+<br>c.[1107delC]             | p.[Asn370Ilefs*22]+<br>p.[Asn370Ilefs*22] | 12.8           | 89             | -1.33         | 21                          | 1.9                                  | 3                              | 155                  | CKD-1                | 22  | Citrate   | Yes                 | No                   | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 2.9                      |     |
| Male   | 2.3                        | ATP6V0A4 | AR          | H     | c.[1185delC]+<br>c.[1185delC]             | p.[Tyr396Thrfs*12]+<br>p.[Tyr396Thrfs*12] | 11.8           | 86             | -2.58         | 22                          | 3.2                                  | 3                              | 143                  | CKD-1                | 19  | Both      | No                  | No                   | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 0.7                      |     |
| Male   | 0                          | ATP6V0A4 | AR          | H     | c.[162_169 dup]+<br>c.[162_169 dup]       | p.[Arg57Lysfs*24]+<br>p.[Arg57Lysfs*24]   | 13.9           | 95.5           | 0.19          | 35                          | 3.2                                  | 3                              | 100                  | CKD-1                | 26  | Citrate   | No                  | Yes                  | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 3.0                      |     |
| Male   | 0.1                        | ATP6V0A4 | AR          | H     | c.[2227C>T]+<br>c.[2227C>T]               | p.[Arg743Trp]+<br>p.[Arg743Trp]           | 12.2           | 91             | -1.14         | 30.9                        | 2.3                                  | 3                              | 107                  | CKD-1                | 25.6                                      | Citrate   |                     |                      | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 2.9                      |     |
| Male   | 0.3                        | ATP6V0A4 | AR          | H     | c.[1181-1185del]+<br>c.[1181-1185del]     | p.[Ala394Valfs*34]+<br>p.[Ala394Valfs*34] | 16.2           | 89.3           | -2.56         | 22                          | 2.5                                  | 3.5                            | 148                  | CKD-1                | 11.6                                      | Citrate   | No                  | No                   | Yes                   | Yes                  | Yes             | Yes             | Yes                                      | Yes                      | 1 Yes                               | 1.6                      | 3.2 |
| Male   | 0.83                       | ATP6V0A4 | AR          | H     | c.[596T>A]+<br>c.[596T>A]                 | p.[Leu199*]+<br>p.[Leu199*]               | 13             | 92             | -2.04         | 46                          | 9.2                                  | 3.7                            | 73.1                 | CKD-2                | 22  | Citrate   | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             | Yes                                      | Yes                      | 1.4 No                              | 2.9                      |     |
| Male   | 0.2                        | ATP6V0A4 | AR          | H     | c.[1107delC]+<br>c.[1107delC]             | p.[Asn370Ilefs*22]+<br>p.[Asn370Ilefs*22] | 14             | 97.4           | -1.31         | 27                          | 6.9                                  | 4                              | 132                  | CKD-1                | 26  | Citrate   | No                  | Yes                  | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 3.8                      |     |
| Female | 0.16                       | ATP6V0A4 | AR          | CH    | c.[1691+2dupT]+<br>c.[c.2293_2294insA]    | p[?]+ p[Ser765Lysfs]                      | 14.6           | 99             | -0.47         | 33                          | 3.3                                  | 4                              | 110                  | CKD-1                | 20  | HCO3      | No                  | No                   | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 3.8                      |     |
| Female | 0                          | ATP6V0A4 | AR          | H     | <b>exact genotype not<br/>retrievable</b> |   | 16.1           | 110.6          | 0.63          | 34                          | 3.4                                  | 5                              | 119                  | CKD-1                | 22.8                                      | Citrate   | No                  | Yes                  | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 5.0                      |     |
| Female | 0.4                        | ATP6V0A4 | AR          | CH    | c.[1185delC]+<br>c.[1691+2dupT]           | p.[Tyr396Thrfs*12]+<br>p.[?]              | 16.5           | 109            | -0.15         | 34                          | 6.7                                  | 5.3                            | 117                  | CKD-1                | 24  | Both      | Yes                 | No                   | Yes                   |                      | Yes             | No              | No                                       | No                       | No                                  | 4.9                      |     |
| Female | 0.16                       | ATP6V0A4 | AR          | CH    | c.[387C>A]+<br>c.[2257C>T]                | p.[Tyr129*]+<br>p.[Gln753*]               | 18.2           | 112            | -0.59         | 30                          | 2.7                                  | 6                              | 136                  | CKD-1                | 24  | Citrate   | Yes                 | No                   | Yes                   | No                   | Yes             | Yes             | Yes                                      | Yes                      | 3.5 No                              | 5.8                      |     |
| Female | 0.2                        | ATP6V0A4 | AR          | CH    | c.[1185delC]+<br>c.[1345C>T]              | p.[Tyr396ThrfsX12]+<br>p.[Arg449Cys]      | 18.4           | 109            | -1.2          | 60                          | 3.1                                  | 6                              | 66                   | CKD-2                | 27  | Both      | No                  | Yes                  | Yes                   | No                   | Yes             | No              | No                                       | No                       | No                                  | 5.8                      |     |
| Male   | 0.4                        | ATP6V0A4 | AR          | H     | c.[1955C>G]+<br>c.[1955C>G]               | p.[Pro652Arg]+<br>p.[Pro652Arg]           | 18.5           | 112            | -0.82         | 32                          | 16.2                                 | 6.11                           | 127.8                | CKD-1                | 20  | Citrate   | No                  | No                   | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 5.7                      |     |
| Male   | 0                          | ATP6V0A4 | AR          | CH    | c.[1029+1G>A]+<br>c.[1345C>T]             | p.[?]+ p.[Arg449Cys]                      | 27.5           | 129            | 0.15          | 55                          | 1.5                                  | 8                              | 86                   | CKD-2                | 20.1                                      | Both      | Yes                 | No                   | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 8.0                      |     |
| Female | 0.2                        | ATP6V0A4 | AR          | H     | c.[1345C>T]+<br>c.[1345C>T]               | p.[Arg449Cys]+<br>p.[Arg449Cys]           | 38.5           | 138.2          | 0.77          | 43                          | 0.8                                  | 9                              | 117                  | CKD-1                | 29  | Citrate   | No                  | Yes                  | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 8.8                      |     |
| Male   | 0.08                       | ATP6V0A4 | AR          | CH    | c.[417_429del]+<br>c.[1920_1921delAA]     | p.[Glu139Aspfs*2]+<br>p.[Ser641Phefs*17]  | 32.8           | 134.2          | -0.74         | 56                          | 2.5                                  | 10                             | 87                   | CKD-2                | 23.1                                      | Citrate   | No                  | Yes                  | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 9.9                      |     |
| Female | 0                          | ATP6V0A4 | AR          | H     | c.[752_755del]+<br>c.[752_755del]         | p.[Glu251Valfs*14]+<br>p.[Glu251Valfs*14] | 39.4           | 142.5          | 0.7           | 47                          | 1.4                                  | 10                             | 111                  | CKD-1                | 23.4                                      | HCO3      | No                  | Yes                  | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 10.0                     |     |
| Female | 0.08                       | ATP6V0A4 | AR          | H     | c.[828_831del]+<br>c.[828_831del]         | p.[Thr277Serfs*25]+<br>p.[Thr277Serfs*25] | 27.2           | 130.3          | -1.25         | 49                          | 1.2                                  | 10                             | 97                   | CKD-1                | 22.1                                      | HCO3      | No                  | Yes                  | Yes                   | Yes                  | Yes             | Yes             | Yes                                      | Yes                      | 7 No                                | 9.9                      |     |
| Male   | 0.25                       | ATP6V0A4 | AR          | H     | c.[2227C>T]+<br>c.[2227C>T]               | p.[Arg743Trp]+<br>p.[Arg743Trp]           | 61.7           | 142            | -0.25         | 66.3                        | 1.2                                  | 11                             | 78                   | CKD-2                | 25.9                                      | Citrate   |                     |                      | Yes                   | No                   | No              | No              | No                                       | No                       | No                                  | 10.8                     |     |

| Gender | Age<br>Diagnosis<br>(year) | Gen      | Inheritance | Trait | c.DNA                                     | Protein result                            | Weight<br>(kg) | Height<br>(cm) | Height<br>SDS | Creatinine<br>(μmol/L) | Alkali<br>serum dose<br>(mEq/kg/day) | Age at<br>last<br>Follow<br>Up | eGFR<br>(ml/<br>min) | CKD<br>Stages<br>I-V | HCO <sub>3</sub> <sup>-</sup><br>(mmol/L) | Treatment | Hyper-<br>calciuria | Metabolic<br>Control | Nephro-<br>calcinosis | Nephro-<br>lithiasis | Hearing<br>loss | Hearing<br>aids | Hearing<br>aids<br>prescription<br>(age) | Cochlear<br>Implantation | Cochlear<br>prescription<br>(years) | Time<br>of FU<br>(years) |      |
|--------|----------------------------|----------|-------------|-------|---|---|----------------|----------------|---------------|------------------------|--------------------------------------|--------------------------------|----------------------|----------------------|---|-----------|---------------------|----------------------|-----------------------|----------------------|-----------------|-----------------|--|--------------------------|-------------------------------------|--------------------------|------|
|        |                            |          |             |       |   |   |                |                |               |                        |                                      |                                |                      |                      |   |           |                     |                      |                       |                      |                 |                 |  |                          |                                     |                          |      |
| Female | 0                          | ATP6V0A4 | AR          | CH    | c.[367G>T]+<br>c.[387C>A]                 | p.[Glu123*]+<br>p.[Tyr129*]               | 37             | 139            | -0.73         | 44                     | 0.9                                  | 11                             | 115                  | CKD-1                | 20  | Both      | No                  | No                   | Yes                   | Yes                  | No              | No              |  | No                       |                                     | 11.0                     |      |
| Female | 0                          | ATP6V0A4 | AR          | H     | <b>exact genotype not<br/>retrievable</b> |   | 30.5           | 148            | 0.51          | 50.3                   | 2                                    | 11                             | 107                  | CKD-1                | 26.6                                      | Citrate   | No                  | Yes                  | Yes                   | No                   | No              | No              |  | No                       |                                     | 11.0                     |      |
| Male   | 0.1                        | ATP6V0A4 | AR          | H     | c.[1312_1315del]+<br>c.[1312_1315del]     | p.[Asp438Metfs*13]+<br>p.[Asp438Metfs*13] | 32.7           | 145.6          | -0.45         | 57                     | 4.6                                  | 12                             | 93                   | CKD-1                | 28  | Citrate   | No                  | Yes                  | Yes                   | No                   | No              | No              |  | No                       |                                     | 11.9                     |      |
| Female | 0.1                        | ATP6V0A4 | AR          | H     | c.[1030-2A>C]+<br>c.[1030-2A>C]           | p.[?]+p.[?]                               | 42             | 149            | -0.8          | 35                     | 5.2                                  | 12.5                           | 155                  | CKD-1                | 22.2                                      | Both      | No                  | Yes                  | Yes                   | No                   | No              | No              |  | No                       |                                     | 12.4                     |      |
| Female | 0.2                        | ATP6V0A4 | AR          | H     | c.[1908+1G>T]+<br>c.[1908+1G>T]           | p.[?]+p.[?]                               | 31             | 140            | -2.48         | 52.2                   | 1.5                                  | 13                             | 98                   | CKD-1                | 25  | Citrate   |                     |                      | Yes                   | No                   | No              | No              |  | No                       |                                     | 12.8                     |      |
| Female | 0.5                        | ATP6V0A4 | AR          | CH    | c.[387C>A]+<br>c.[2257C>T]                | p.[Tyr129*]+<br>p.[Gln753*]               | 45.5           | 158            | -0.38         | 43                     | 1.2                                  | 14                             | 134                  | CKD-1                | 26  | Citrate   | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 2.8                      | Yes                                 | 10.8                     | 13.5 |
| Male   | 0                          | ATP6V0A4 | AR          | H     | c.[752_755del]+<br>c.[752_755del]         | p.[Glu251Valfs*14]+<br>p.[Glu251Valfs*14] | 46.5           | 154.6          | -1.12         | 68                     | 1.6                                  | 14                             | 83                   | CKD-2                | 19.3                                      | HCO3      | No                  | No                   | Yes                   | No                   | No              | No              |  | No                       |                                     | 14.0                     |      |
| Female | 2.5                        | ATP6V0A4 | AR          | H     | c.[322C>T]+<br>c.[322C>T]                 | p.[Gln108*]+<br>p.[Gln108*]               | 61             | 164.5          | 0.56          | 49                     | 0.7                                  | 14.5                           | 123                  | CKD-1                | 23  | HCO3      | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 12.5                     | Yes                                 | 13                       | 12.0 |
| Male   | 0                          | ATP6V0A4 | AR          | CH    | c.[25_27del]+<br>c.[863C>A]               | p.[Glu9del]+<br>p.[Ala288Asp]             | 62             | 173            | 0.37          | 68                     | 1                                    | 15                             | 93                   | CKD-1                | 28  | Citrate   | No                  | Yes                  | Yes                   | No                   | No              | No              |  | No                       |                                     | 15.0                     |      |
| Male   | 0.5                        | ATP6V0A4 | AR          | CH    | c.[1478+1G>A]+<br>c.[2190C>G]             | p.[?]+<br>p.[Tyr730*]                     | 64.4           | 174            | 0.36          | 68                     | 0.9                                  | 15.3                           | 93                   | CKD-1                | 23.8                                      | Both      | No                  | Yes                  | Yes                   | No                   | No              | No              |  | No                       |                                     | 14.8                     |      |
| Male   | 0.1                        | ATP6V0A4 | AR          | H     | c.[1691+1G>A]+<br>c.[1691+1G>A]           | p.[?]+p.[?]                               | 60.6           | 174.4          | 0.05          | 93                     | 2.5                                  | 16                             | 68                   | CKD-2                | 26  | HCO3      | No                  | Yes                  | Yes                   | No                   | No              | No              |  | No                       |                                     | 15.9                     |      |
| Male   | 0.3                        | ATP6V0A4 | AR          | CH    | c.[16C>T]+<br>c.[970delG]                 | p.[Arg6*]+<br>p.[Glu324Argfs*22]          | 69.6           | 180.2          | 0.87          | 93                     | 1                                    | 16                             | 71                   | CKD-2                | 27  | HCO3      | No                  | Yes                  | Yes                   | No                   | No              | No              |  | No                       |                                     | 15.7                     |      |
| Female | 0.1                        | ATP6V0A4 | AR          | H     | c.[188_189del]+<br>c.[188_189del]         | p.[Arg63Asnfs*12]+<br>p.[Arg63Asnfs*12]   | 46.5           | 155            | -1.17         | 63                     | 2.3                                  | 16                             | 90                   | CKD-1                | 24  | HCO3      | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 14                       | No                                  |                          | 15.9 |
| Female | 0                          | ATP6V0A4 | AR          | CH    | c.[367G>T]+<br>c.[387C>A]                 | p.[Glu123*]+<br>p.[Tyr129*]               | 54             | 169            | 0.99          | 44                     | 1.8                                  | 16                             | 140                  | CKD-1                | 25  | Both      | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 12                       | No                                  |                          | 16.0 |
| Male   | 1                          | ATP6V0A4 | AR          | H     | c.[1920_1921delAA]+<br>c.[1920_1921delAA] | p.[Ser641Phefs*17]+<br>p.[Ser641Phefs*17] | 80             | 166            | -1.26         | 127                    | 1.1                                  | 17                             | 48                   | CKD-3                | 20.8                                      | HCO3      | No                  | No                   | Yes                   | No                   | No              | No              |  | No                       |                                     | 16.0                     |      |
| Male   | 0                          | ATP6V0A4 | AR          | CH    | c.[25_27del]+<br>c.[863C>A]               | p.[Glu9del]+<br>p.[Ala288Asp]             | 82             | 187            | 1.64          | 88                     | 0.7                                  | 17                             | 78                   | CKD-2                | 32  | Citrate   | No                  | Yes                  | Yes                   | No                   | No              | No              |  | No                       |                                     | 17.0                     |      |
| Male   | 0.1                        | ATP6V0A4 | AR          | H     | c.[418-1G>A]+<br>1G>A]                    | c.[418-<br>p.[?]+p.[?]                    | 41.5           | 148.2          | -3.55         | 226                    | 1.7                                  | 17                             | 24                   | CKD-4                | 21  | HCO3      | Yes                 | No                   | No                    | No                   | No              | No              |  | No                       |                                     | 16.9                     |      |
| Male   | 0.1                        | ATP6V0A4 | AR          | H     | c.[1345C>T]+<br>c.[1345C>T]               | p.[Arg449Cys]+<br>p.[Arg449Cys]           | 67.5           | 158            | -2.36         | 72                     | 2.4                                  | 17.3                           | 80                   | CKD-2                | 22.2                                      | Both      | No                  | Yes                  | Yes                   | No                   | No              | No              |  | No                       |                                     | 17.2                     |      |
| Male   | 0.25                       | ATP6V0A4 | AR          | CH    | c.[387C>A]+<br>c.[2257C>T]                | p.[Tyr129*]+<br>p.[Gln753*]               | 42             | 160            | -2.22         | 50                     | 1.3                                  | 18                             | 116.8                | CKD-1                | 24  | Citrate   | No                  | Yes                  | Yes                   |                      | No              | No              |  | No                       |                                     | 17.8                     |      |
| Female | 0.3                        | ATP6V0A4 | AR          | CH    | c.[1185delC]+<br>c.[1572G>A]              | p.[Tyr396Thrfs*12]+<br>p.[Pro524=]        | 60.4           | 165            | 0.26          | 73                     | 2                                    | 23                             | 85.7                 | CKD-2                | 17.4                                      | Both      | No                  | No                   | Yes                   | No                   | No              | No              |  | No                       |                                     | 22.7                     |      |
| Female | 0.25                       | ATP6V0A4 | AR          | CH    | c.[414_417+10del14]<br>+ c.[1571 C>T]     | p.[?]+<br>p.[Pro524leu]                   | 60             | 159            | -0.67         | 86.6                   | 1.2                                  | 27                             | 68.1                 | CKD-2                | 15.9                                      | Both      | No                  | No                   | Yes                   | Yes                  | Yes             | Yes             |  | No                       |                                     | 26.8                     |      |
| Female | 0.08                       | ATP6V0A4 | AR          | H     | c.[2137delG]+<br>c.[2137delG]             | p.[Glu713Serfs*50]+<br>p.[Glu713Serfs*50] | 62.4           | 162            | -0.21         | 48                     | 1.7                                  | 28                             | 133.6                | CKD-1                | 19  | Both      | No                  | No                   | Yes                   | Yes                  | Yes             | Yes             |  | 7                        | No                                  |                          | 27.9 |
| Female | 0.08                       | ATP6V0A4 | AR          | H     | c.[322C>T]+<br>c.[322C>T]                 | p.[Gln108*]+<br>p.[Gln108*]               | 59.7           | 151            | -1.9          | 61                     | 1.5                                  | 29                             | 100.6                | CKD-1                | 24  | HCO3      | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 4                        | No                                  |                          | 28.9 |
| Female | 2                          | ATP6V0A4 | AR          | H     | c.[1345C>T]+<br>c.[1345>T]                | p.[Arg449Cys]+<br>p.[Arg449Cys]           | 47             | 160            | -0.52         | 148                    | 1                                    | 32                             | 35.5                 | CKD-3                | 17.8                                      | Citrate   |                     |                      |                       | Yes                  | Yes             | No              |  | No                       |                                     | 30.0                     |      |
| Female | 0.6                        | ATP6V0A4 | AR          | H     | c.[1345C>T]+<br>c.[1345>T]                | p.[Arg449Cys]+<br>p.[Arg449Cys]           | 54             | 158            | -0.82         | 105                    | 1.7                                  | 32                             | 52.7                 | CKD-3                | 16.5                                      | Citrate   | No                  | No                   | Yes                   | No                   | Yes             | Yes             |  | 4                        | No                                  |                          | 31.4 |
| Female | 0.1                        | ATP6V0A4 | AR          | H     | c.[2426A>C]+<br>c.[2426A>C]               | p.[Hys809Arg]+<br>p.[Hys809Arg]           | 68             | 162            | -0.21         | 61.9                   | 1.3                                  | 35                             | 95.2                 | CKD-1                | 22.6                                      | Both      | No                  | Yes                  | Yes                   | Yes                  | Yes             | Yes             |  | No                       |                                     | 34.9                     |      |
| Male   | 0.08                       | ATP6V0A4 | AR          | H     | c.[417+1G>A]+<br>c.[417+1G>A]             | p.[?]+p.[?]                               | 61.8           | 160.5          | -0.3          | 124                    | 0.8                                  | 38                             | 56.6                 | CKD-3                | 24.5                                      | HCO3      | No                  | Yes                  | Yes                   | No                   | Yes             | No              |  | No                       |                                     | 37.9                     |      |
| Female | 0.25                       | ATP6V0A4 | AR          | H     | c.[196+4A>G]+<br>c.[196+4A>G]             | p.[?]+p.[?]                               | 73             | 165            | 0.26          | 74.3                   | 0.7                                  | 39                             | 75.4                 | CKD-2                | 17.4                                      | Both      |                     |                      | Yes                   | Yes                  | Yes             | Yes             |  | No                       |                                     | 38.8                     |      |
| Male   | 0.3                        | ATP6V0A4 | AR          | H     | c.[2215G>C]+<br>c.[2215G>C]               | p.[Ala739Pro]+<br>p.[Ala739Pro]           | 92             | 189            | 1.72          | 98                     | 1                                    | 39                             | 73.9                 | CKD-2                | 27  | Both      | No                  | Yes                  | Yes                   | Yes                  | Yes             | No              |  | No                       |                                     | 38.7                     |      |

| Gender | Age<br>Diagnosis<br>(year) | Gen      | Inheritance | Trait | c.DNA   | Protein result  | Weight<br>(kg) | Height<br>(cm) | Height<br>SDS | Creatinine<br>(μmol/L) | Alkali<br>serum dose<br>(mEq/<br>kg/day) | Age at<br>last<br>Follow<br>Up | eGFR<br>(ml/<br>min) | CKD<br>Stages<br>I-V | HCO <sub>3</sub> <sup>-</sup><br>(mmol/L) | Treatment | Hyper-<br>calciuria | Metabolic<br>Control | Nephro-<br>calcinosis | Nephro-<br>lithiasis | Hearing<br>loss | Hearing<br>aids | Hearing<br>aids<br>prescription<br>(age) | Cochlear<br>Implantation | Cochlear<br>prescription<br>(years) | Time<br>of FU<br>(years) |      |
|--------|----------------------------|----------|-------------|-------|---|---|----------------|----------------|---------------|------------------------|--|--------------------------------|----------------------|----------------------|---|-----------|---------------------|----------------------|-----------------------|----------------------|-----------------|-----------------|--|--------------------------|-------------------------------------|--------------------------|------|
|        |                            |          |             |       |   |   |                |                |               |                        |  |                                |                      |                      |   |           |                     |                      |                       |                      |                 |                 |  |                          |                                     |                          |      |
| Female | 0                          | ATP6V0A4 | AR          | H     | c.[752_755del]+<br>c.[752_755del]<br>c.[40C>T]+<br>c.[816+1G>A]+          | p.[Glu251Valfs*14]+<br>p.[Glu251Valfs*14]<br>p.[Gln14*]+<br>p.[Pro631Ser]               | 54.6           | 156.1          | -1.13         | 68                     | 1.7                                      | 39                             | 83.6                 | CKD-2                | 30.5                                      | HCO3      | No                  | Yes                  | Yes                   | Yes                  | No              | No              |  |                          | No                                  |                          | 39.0 |
| Male   | 0.08                       | ATP6V0A4 | AR          | CH    | c.[1891C>T]<br>c.[1185delC]+<br>c.[1754_1781dup28]<br>c.[1356del]+        | p.[Tyr396Thrfs*12]+<br>p.[Ile594Metfs*18]<br>p.[Phe452fsX35]+<br>p.[Phe452fsX35]+       | 52             | 169            | 0.89          | 97                     | 1.2                                      | 40                             | 74.4                 | CKD-2                | 29  | HCO3      | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 20                       | No                                  |                          | 39.9 |
| Female | 0.1                        | ATP6V0A4 | AR          | CH    | c.[1356del]+<br>c.[91C>T]+<br>c.[1155duplC]<br>c.[175-1G>C]+              | p.[Phe452fsX35]+<br>p.[Phe452fsX35]<br>p.[Ile386Hisfs*56]<br>p.[?]+                     | 54             | 146            | -2.66         | 90                     | 1.5                                      | 43                             | 59.3                 | CKD-3                | 23.8                                      | Both      | No                  | Yes                  | Yes                   | Yes                  | Yes             | Yes             |  | 39                       | No                                  |                          | 42.9 |
| Male   | 0                          | ATP6V1B1 | AR          | H     | c.[1356del]<br>c.[91C>T]+<br>c.[1155duplC]<br>c.[175-1G>C]+               | p.[Phe452fsX35]<br>p.[Arg31*]+<br>p.[Ile386Hisfs*56]<br>p.[?]+                          | 6.9            | 65             | -1.15         | 17.7                   | 3.6                                      | 0.5                            | 134                  | CKD-1                | 25  | Citrate   | No                  | Yes                  | Yes                   | No                   | Yes             | No              |  |                          | No                                  |                          | 0.5  |
| Female | 0                          | ATP6V1B1 | AR          | CH    | c.[1155duplC]<br>c.[175-1G>C]+<br>c.[1155duplC]<br>c.[91C>T]+             | p.[Ile386Hisfs*56]<br>p.[?]+<br>p.[Ile386Hisfs*56]<br>p.[Arg31*]+                       | 5.6            | 66             | -0.05         | 16                     | 1.8                                      | 0.5                            | 151                  | CKD-1                | 22  | Citrate   | No                  | Yes                  | Yes                   | No                   |                 | No              |  |                          | No                                  |                          | 0.5  |
| Female | 1.4                        | ATP6V1B1 | AR          | CH    | c.[1155duplC]<br>c.[91C>T]+<br>c.[91C>T]<br>c.[175-1G>C]+                 | p.[Ile386Hisfs*56]<br>p.[Arg31*]+<br>p.[Arg31*]<br>p.[?]+                               | 11.3           | 83.5           | -1.3          | 32                     | 1.4                                      | 2.4                            | 95                   | CKD-1                | 18.1                                      | HCO3      | No                  | No                   | Yes                   | No                   | Yes             | Yes             |  | 1.9                      | No                                  |                          | 1.0  |
| Female | 1.2                        | ATP6V1B1 | AR          | H     | c.[91C>T]<br>c.[91C>T]<br>c.[175-1G>C]+<br>c.[1155duplC]                  | p.[Arg31*]<br>p.[Arg31*]<br>p.[?]+<br>p.[Ile386Hisfs*56]                                | 9.5            | 70             | -5.52         | 36                     | 9.5                                      | 2.5                            | 71.0                 | CKD-2                | 20  | Citrate   | Yes                 | No                   | Yes                   | No                   | Yes             | Yes             |  | 1.2                      | No                                  |                          | 1.3  |
| Female | 2.5                        | ATP6V1B1 | AR          | CH    | c.[1155duplC]<br>c.[232G>A]+<br>c.[988G>C]<br>c.[1387G>T]+                | p.[Ile386Hisfs*56]<br>p.[Gly78Arg]+<br>p.[Glu330Gln]<br>p.[Glu463*]+                    | 15.7           | 94             | -0.89         | 44                     | 2.4                                      | 3.5                            | 78                   | CKD-2                | 16.3                                      | Both      | No                  | No                   | Yes                   | No                   | Yes             | No              |  |                          | No                                  |                          | 1.0  |
| Female | 2                          | ATP6V1B1 | AR          | CH    | c.[988G>C]<br>c.[1387G>T]+<br>c.[1387G>T]<br>c.[242T>C]+                  | p.[Glu330Gln]<br>p.[Glu463*]+<br>p.[Glu463*]<br>p.[Leu81Pro]+                           | 13.8           | 92.3           | -1.38         | 35                     | 1.1                                      | 3.5                            | 96                   | CKD-1                | 18  | HCO3      | No                  | No                   | Yes                   | No                   | Yes             | Yes             |  | 0.5                      | No                                  |                          | 1.5  |
| Female | 0.3                        | ATP6V1B1 | AR          | H     | c.[1387G>T]<br>c.[242T>C]+<br>c.[242T>C]<br>c.[91C>T]+                    | p.[Glu463*]<br>p.[Glu463*]<br>p.[Leu81Pro]<br>p.[Leu81Pro]                              | 19.2           | 98.2           | -0.7          | 25                     | 0.9                                      | 4                              | 143                  | CKD-1                | 21  | Citrate   | No                  | No                   | Yes                   | No                   | Yes             | No              |  |                          | No                                  |                          | 3.7  |
| Female | 2                          | ATP6V1B1 | AR          | H     | c.[242T>C]<br>c.[91C>T]+<br>c.[91C>T]<br>c.[1037C>G]+                     | p.[Leu81Pro]<br>p.[Arg31*]+<br>p.[Arg31*]<br>p.[Pro346Arg]+                             | 20.5           | 100            | -0.24         | 35.4                   | 1.7                                      | 4                              | 103                  | CKD-1                | 25  | Both      | No                  | Yes                  | Yes                   | No                   | No              | No              |  |                          | No                                  |                          | 2.0  |
| Female | 0.5                        | ATP6V1B1 | AR          | H     | c.[91C>T]<br>c.[1037C>G]+<br>c.[1037C>G]<br>c.[242T>C]+                   | p.[Arg31*]<br>p.[Arg31*]<br>p.[Pro346Arg]<br>p.[Leu81Pro]+                              | 18             | 105            | 0.89          | 30.9                   | 2  | 4                              | 124                  | CKD-1                | 22.3                                      | Citrate   | Yes                 | No                   | Yes                   | No                   | Yes             | Yes             |  | 1                        | No                                  |                          | 3.5  |
| Male   | 0.3                        | ATP6V1B1 | AR          | H     | c.[1037C>G]<br>c.[242T>C]+<br>c.[242T>C]<br>c.[242T>C]+                   | p.[Pro346Arg]<br>p.[Leu81Pro]<br>p.[Leu81Pro]<br>p.[Leu81Pro]+                          | 30.3           | 130.5          | 4.7           | 53                     | 3  | 5                              | 90                   | CKD-1                | 26  | Citrate   | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 3                        | No                                  |                          | 4.7  |
| Male   | 0.2                        | ATP6V1B1 | AR          | H     | c.[242T>C]<br>c.[242T>C]<br>c.[242T>C]<br>c.[332del]                      | p.[Leu81Pro]<br>p.[Leu81Pro]<br>p.[Leu81Pro]<br>p.[Asp111Alafs*53]                      | 16             | 103            | -1.32         | 35                     | 2.8                                      | 5                              | 107                  | CKD-1                | 24.9                                      | Both      | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 2.5                      | No                                  |                          | 4.8  |
| Male   | 0.4                        | ATP6V1B1 | AR          | CH    | c.[332del]<br>c.[1148_1149insC]+<br>c.[1148_1149insC]<br>c.[1249-3C>G]+   | p.[Asp111Alafs*53]<br>p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56]<br>p.[?]+p.[?]          | 24             | 110            | 0.18          | 45                     | 2.3                                      | 5                              | 89                   | CKD-2                | 22.3                                      | Both      | No                  | Yes                  | Yes                   | No                   | No              | No              |  |                          | No                                  |                          | 4.6  |
| Female | 0.5                        | ATP6V1B1 | AR          | H     | c.[1148_1149insC]<br>c.[1148_1149insC]<br>c.[1249-3C>G]+<br>c.[1249-3C>G] | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56]<br>p.[?]+p.[?]<br>p.[Ile386Hisfs*56]+         | 16.5           | 105.6          | -1.37         | 39                     | 4.2                                      | 5.7                            | 99                   | CKD-1                | 17  | Both      | No                  | No                   | Yes                   | No                   | Yes             | Yes             |  | 2.5                      | Yes                                 |                          | 4.9  |
| Male   |                            | ATP6V1B1 | AR          | H     | c.[1249-3C>G]<br>c.[1148_1149insC]<br>c.[1148_1149insC]<br>c.[91C>T]+     | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56]<br>p.[Ile386Hisfs*56]<br>p.[Arg31*]+          | 25.8           | 116            | 0.39          | 30                     | 1.9                                      | 5.8                            | 141                  | CKD-1                | 22  | Both      | No                  | Yes                  | No                    | Yes                  | No              | No              |  |                          | No                                  |                          |      |
| Male   | 2.5                        | ATP6V1B1 | AR          | H     | c.[1148_1149insC]<br>c.[1148_1149insC]<br>c.[91C>T]+<br>c.[91C>T]         | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56]<br>p.[Arg31*]+<br>p.[Arg31*]                  | 16.9           | 106.8          | -1.71         | 25                     | 1.9                                      | 6                              | 156                  | CKD-1                | 25  | Both      | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 2.5                      | Yes                                 |                          | 5    |
| Female | 0                          | ATP6V1B1 | AR          | H     | c.[91C>T]<br>c.[1155dupC]+<br>c.[1155dupC]<br>c.[1155dupC]+               | p.[Arg31*]<br>p.[Arg31*]<br>p.[Ile386Hisfs*56]<br>p.[Ile386Hisfs*56]                    | 21             | 114.4          | -0.2          | 32                     | 1  | 6                              | 130                  | CKD-1                | 24.2                                      | Citrate   | No                  | Yes                  | Yes                   | No                   | Yes             | No              |  |                          | Yes                                 |                          | 0.7  |
| Female | 0                          | ATP6V1B1 | AR          | H     | c.[1155dupC]<br>c.[1155dupC]<br>c.[1155dupC]+<br>c.[1155dupC]             | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56]<br>p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56]  | 30.5           | 122            | 0.55          | 35                     | 1  | 6.6                            | 127                  | CKD-1                | 22  | Both      | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  |                          | Yes                                 |                          | 5.8  |
| Male   | 0.6                        | ATP6V1B1 | AR          | H     | c.[1155dupC]<br>C.[1156_1157insC]+<br>c.[1354delT]<br>c.[1249-3C>G]+      | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56]<br>p.[Ile386Thrfs*56]+ | 23             | 119.8          | -0.38         | 43                     | 2.1                                      | 7                              | 102                  | CKD-1                | 26  | HCO3      | No                  | Yes                  | No                    |                      | Yes             | Yes             |  |                          | Yes                                 |                          | 6.4  |
| Male   | 0.3                        | ATP6V1B1 | AR          | CH    | c.[1156_1157insC]<br>c.[1354delT]<br>c.[1249-3C>G]+<br>c.[1249-3C>G]      | p.[Ile386Thrfs*56]+<br>p.[Phe452Leufs*35]<br>p.[?]+[?]<br>p.[?]+[?]                     | 26             | 124            | 0.36          | 26.5                   | 2.1                                      | 7                              | 171                  | CKD-1                | 26  | Citrate   | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 1                        | No                                  |                          | 6.7  |
| Male   | 0.6                        | ATP6V1B1 | AR          | H     | c.[1249-3C>G]<br>c.[340C>T]+<br>c.[785+1G>A]<br>c.[815C>G]+               | p.[?]+[?]<br>p.[Arg114*]+p.[?]<br>p.[Arg272Gly]+<br>p.[Asp352Hys]                       | 23             | 124            | 0.36          | 34                     | 1.7                                      | 7                              | 133                  | CKD-1                | 25  | HCO3      | No                  | Yes                  | Yes                   | No                   | No              | No              |  |                          | No                                  |                          | 6.4  |
| Female | 1                          | ATP6V1B1 | AR          | CH    | c.[785+1G>A]<br>c.[815C>G]+<br>c.[1054A>C]<br>c.[1037C>G]+                | p.[Arg114*]+p.[?]<br>p.[Arg272Gly]+<br>p.[Asp352Hys]<br>p.[Pro346Arg]+                  | 24.3           | 126.5          | 0.92          | 46                     | 3.3                                      | 7                              | 100                  | CKD-1                | 25  | Citrate   | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 2                        | No                                  |                          | 6.0  |
| Male   | 0.16                       | ATP6V1B1 | AR          | CH    | c.[1054A>C]<br>c.[1037C>G]+<br>c.[1397C>A]<br>c.[490_499del]+             | p.[Pro346Arg]+<br>p.[Ser466*]<br>p.[Ile164Alafs*8]+<br>p.[Ile164Alafs*8]                | 23.5           | 118            | -0.75         | 90                     | 0.3                                      | 7                              | 48                   | CKD-3                | 26.5                                      | Citrate   | No                  | Yes                  | Yes                   | No                   | No              | No              |  |                          | No                                  |                          | 6.8  |
| Male   | 0.3                        | ATP6V1B1 | AR          | CH    | c.[1397C>A]<br>c.[490_499del]<br>c.[490_499del]<br>c.[242T>C]+            | p.[Ser466*]<br>p.[Ile164Alafs*8]+<br>p.[Ile164Alafs*8]<br>p.[Leu81Pro]+                 | 39             | 135            | 1.74          | 40                     | 2.6                                      | 7.5                            | 123                  | CKD-1                | 23.8                                      | Both      | No                  | Yes                  | No                    | No                   | Yes             | Yes             |  |                          | No                                  |                          | 7.2  |
| Female | 0.1                        | ATP6V1B1 | AR          | H     | c.[490_499del]<br>c.[490_499del]<br>c.[242T>C]+<br>c.[1155duplC]          | p.[Ile164Alafs*8]<br>p.[Ile164Alafs*8]<br>p.[Leu81Pro]<br>p.[Ile386Hisfs*56]            | 28.6           | 130.3          | -0.51         | 45                     | 3.8                                      | 9                              | 106                  | CKD-1                | 25  | Citrate   | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 0.5                      | Yes                                 |                          | 2    |
| Male   | 1                          | ATP6V1B1 | AR          | CH    | c.[1155duplC]<br>c.[1155duplC]  | p.[Leu81Pro]<br>p.[Ile386Hisfs*56]  | 35.6           | 138.6          | 0.03          | 41                     | 2.2                                      | 10                             | 123                  | CKD-1                | 22.5                                      | Citrate   | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  |                          | Yes                                 |                          | 11.3 |

| Gender | Age<br>Diagnosis<br>(year) | Gen      | Inheritance | Trait | c.DNA                                     | Protein result                            | Weight<br>(kg) | Height<br>(cm) | Height<br>SDS | Creatinine<br>(μmol/L) | Alkali                         | Age at               | eGFR<br>(ml/<br>min) | CKD<br>Stages<br>I-V | HCO <sub>3</sub> <sup>-</sup><br>(mmol/L) | Treatment | Hyper-<br>calciuria | Metabolic<br>Control | Nephro-<br>calcinosis | Nephro-<br>lithiasis | Hearing<br>loss | Hearing<br>aids | Hearing<br>aids<br>prescription<br>(age) | Cochlear<br>Implantation | Cochlear<br>prescription<br>(years) | Time<br>of FU<br>(years) |      |      |
|--------|----------------------------|----------|-------------|-------|---|---|----------------|----------------|---------------|------------------------|--------------------------------|----------------------|----------------------|----------------------|---|-----------|---------------------|----------------------|-----------------------|----------------------|-----------------|-----------------|--|--------------------------|-------------------------------------|--------------------------|------|------|
|        |                            |          |             |       |   |   |                |                |               |                        | serum dose<br>(mEq/<br>kg/day) | last<br>Follow<br>Up |                      |                      |   |           |                     |                      |                       |                      |                 |                 |  |                          |                                     |                          |      |      |
| Female | 0                          | ATP6V1B1 | AR          | H     | c.[242T>C]+<br>c.[242T>C]                 | p.[Leu81Pro]+<br>p.[Leu81Pro]             | 26             | 128            | -1.56         | 44                     | 2.3                            | 10                   | 106                  | CKD-1                | 22  | HCO3      |                     |                      | Yes                   | No                   | Yes             | Yes             |  | 0                        | Yes                                 |                          | 2    | 10.0 |
| Female | 0                          | ATP6V1B1 | AR          | H     | c.[1155dupC]+<br>c.[1155dupC]             | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56] | 25             | 138            | -0.86         | 35                     | 1                              | 11                   | 144                  | CKD-1                | 21  | Both      | No                  | No                   | Yes                   | Yes                  | Yes             | Yes             |  | 3                        | No                                  |                          |      | 11.0 |
| Male   | 0.5                        | ATP6V1B1 | AR          | H     | c.[242T>C]+<br>c.[242T>C]                 | p.[Leu81Pro]+<br>p.[Leu81Pro]             | 35             | 140            | -0.53         | 55                     | 2.9                            | 11                   | 93                   | CKD-1                | 23  | Both      | No                  | Yes                  | Yes                   | No                   |                 |                 |  |                          |                                     |                          |      | 10.5 |
| Male   | 0.1                        | ATP6V1B1 | AR          | H     | c.[1155dupC]+<br>c.[1155dupC]             | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56] | 23             | 121            | -3.69         | 53                     | 3.7                            | 11.5                 | 83                   | CKD-2                | 16  | Citrate   | Yes                 | No                   | Yes                   | No                   | Yes             | Yes             |  | 5                        | No                                  |                          |      | 11.4 |
| Female | 5                          | ATP6V1B1 | AR          | CH    | c.[340C>T]+<br>c.[1155dupC]               | p.[Arg114*]+<br>p.[Ile386Hisfs*56]        | 44.5           | 151.5          | 0.56          | 57                     | 1.2                            | 11.5                 | 97                   | CKD-1                | 18  | Citrate   | No                  | No                   | Yes                   | No                   | Yes             | Yes             |  | 5                        | Yes                                 |                          | 11.4 | 6.5  |
| Male   | 0.5                        | ATP6V1B1 | AR          | CH    | c.[91C>T]+<br>c.[497delC]                 | p.[Arg31*]+<br>p.[Thr166Argfs*9]          | 31.4           | 136            | -1.83         | 41.5                   | 1.9                            | 12                   | 120                  | CKD-1                | 22.7                                      | Citrate   | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 2                        | No                                  |                          |      | 11.5 |
| Male   | 0.5                        | ATP6V1B1 | AR          | H     | c.[1155dupC]+<br>c.[1155dupC]             | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56] | 26             | 127            | -3.49         | 70.7                   | 2.6                            | 12.5                 | 66                   | CKD-2                | 17  | Citrate   | No                  | No                   | Yes                   | No                   | Yes             | Yes             |  | 6                        | No                                  |                          |      | 12.0 |
| Male   | 0                          | ATP6V1B1 | AR          | H     | c.[135-1G>C]+<br>c.[135-1G>C]             | p.[?]+p.[?]                               | 48             | 156            | -0.05         | 53                     | 1.1                            | 13                   | 107                  | CKD-1                | 23  | Citrate   | No                  | Yes                  | Yes                   | Yes                  | Yes             | Yes             |  | 1                        | No                                  |                          |      | 13.0 |
| Male   | 2                          | ATP6V1B1 | AR          | CH    | c.[905G>C]+<br>c.[1469C>T]                | p.[Arg302Pro]+<br>p.[Pro490Leu]           | 60.4           | 151.6          | -0.56         | 66                     | 2                              | 13                   | 84                   | CKD-2                | 18  | Citrate   | Yes                 | No                   | Yes                   | No                   | Yes             | No              |  |                          | No                                  |                          |      | 11.0 |
| Male   | 0.5                        | ATP6V1B1 | AR          | H     | <b>exact genotype not<br/>retrievable</b> |   | 48.7           | 152            | -0.56         | 40.7                   | 1.2                            | 13                   | 136                  | CKD-1                | 26  | Citrate   | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 2                        | Yes                                 |                          | 12   | 12.5 |
| Male   | 7.3                        | ATP6V1B1 | AR          | H     | c.[242T>C]+<br>c.[242T>C]                 | p.[Leu81Pro]+<br>p.[Leu81Pro]             | 49.9           | 157            | -0.65         | 70.7                   | 1.8                            | 13.8                 | 81                   | CKD-2                | 22.7                                      | Both      | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 10                       | No                                  |                          |      | 6.5  |
| Male   | 2                          | ATP6V1B1 | AR          | H     | c.[1155dupC]+<br>c.[1155dupC]             | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56] | 39.7           | 150.5          | -1.72         | 62                     | 0.4                            | 14.2                 | 89                   | CKD-2                | 22  | Citrate   | No                  | Yes                  | Yes                   |                      | Yes             | Yes             |  |                          | Yes                                 |                          |      | 12.2 |
| Male   |                            | ATP6V1B1 | AR          | H     | c.[1249-3C>G]+<br>c.[1249-3C>G]           | p.[?]+p.[?]                               | 60.4           | 159            | -1.16         | 53                     | 1.1                            | 14.7                 | 110                  | CKD-1                | 21  | Both      | No                  | No                   | No                    | Yes                  | No              | No              |  |                          | No                                  |                          |      | No   |
| Male   | 3                          | ATP6V1B1 | AR          | H     | c.[242T>C]+<br>c.[242T>C]                 | p.[Leu81Pro]+<br>p.[Leu81Pro]             | 71             | 166            | -0.99         | 97                     | 1.3                            | 16                   | 62                   | CKD-2                | 21  | Both      | No                  | No                   | Yes                   | No                   | Yes             | No              |  |                          | No                                  |                          |      | 13.0 |
| Male   | 0                          | ATP6V1B1 | AR          | H     | c.[469T>C]+<br>c.[469T>C]                 | p.[Arg157Cys]+<br>p.[Arg157Cys]           | 52             | 163            | -1.37         | 74                     | 0.7                            | 16                   | 80                   | CKD-2                | 24  | HCO3      | No                  | Yes                  | Yes                   | No                   | No              | No              |  |                          | No                                  |                          |      | 16.0 |
| Male   | 0.08                       | ATP6V1B1 | AR          | H     | c.[585+1G>A]+<br>c.[585+1G>A]             | p.[?]+p.[?]                               | 37             | 145            | -3.48         | 57                     | 1.1                            | 16                   | 93                   | CKD-1                | 21  | Citrate   | No                  | No                   | Yes                   | Yes                  | Yes             | Yes             |  | 6                        | Yes                                 |                          | 23   | 15.9 |
| Female | 6                          | ATP6V1B1 | AR          | H     | c.[175-1G>C]+<br>c.[175-1G>C]             | p.[?]+p.[?]                               | 52             | 170.5          | 1.28          | 62                     | 1.2                            | 16.3                 | 100                  | CKD-1                | 17  | Both      | No                  | No                   | Yes                   | Yes                  | Yes             | Yes             |  | 4                        | Yes                                 |                          | 16.6 | 10.3 |
| Male   | 0.5                        | ATP6V1B1 | AR          | CH    | c.[91C>T]+<br>c.[497delC]                 | p.[Arg31*]+<br>p.[Thr166Argfs*9]          | 51.9           | 152            | -2.89         | 85.7                   | 1.9                            | 16.5                 | 65                   | CKD-2                | 20.4                                      | Citrate   | Yes                 | No                   | Yes                   | No                   | Yes             | Yes             |  | 3                        | No                                  |                          |      | 16.0 |
| Male   | 0.1                        | ATP6V1B1 | AR          | H     | c.[1037C>G]+<br>c.[1037C>G]               | p.[Pro346Arg]+<br>p.[Pro346Arg]           | 71             | 168.2          | -1.13         | 89.2                   | 0.5                            | 18                   | 68.8                 | CKD-2                | 26  | Both      | No                  | Yes                  | No                    | Yes                  | Yes             | Yes             |  | 3                        | Yes                                 |                          | 16   | 17.9 |
| Female | 0.11                       | ATP6V1B1 | AR          | H     | c.[1155dupC]+<br>c.[1155dupC]             | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56] | 47.5           | 166.8          | 0.6           | 77                     | 2.9                            | 18                   | 79.1                 | CKD-2                | 24.1                                      | Citrate   | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 1                        | No                                  |                          |      | 17.9 |
| Male   | 4.5                        | ATP6V1B1 | AR          | H     | c.[242T>C]+<br>c.[242T>C]                 | p.[Leu81Pro]+<br>p.[Leu81Pro]             | 66.5           | 157            | -2.62         | 136                    | 1.7                            | 18                   | 42.1                 | CKD-3                | 16  | Both      | Yes                 | No                   | Yes                   | No                   | Yes             | No              |  |                          | No                                  |                          |      | 13.5 |
| Female | 1                          | ATP6V1B1 | AR          | H     | <b>exact genotype not<br/>retrievable</b> |   | 58.1           | 157.9          | -0.79         | 60                     | 0.6                            | 18                   | 96.1                 | CKD-1                | 25.6                                      | HCO3      | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 3.2                      | No                                  |                          |      | 17.0 |
| Male   | 4                          | ATP6V1B1 | AR          | H     | c.[1037C>G]+<br>c.[1037C>G]               | p.[Pro346Arg]+<br>p.[Pro346Arg]           | 70             | 182            | 0.76          | 80                     | 1                              | 19                   | 83                   | CKD-2                | 24.5                                      | HCO3      | No                  | Yes                  | Yes                   | No                   | Yes             | No              |  |                          | No                                  |                          |      | 15.0 |
| Female | 0.16                       | ATP6V1B1 | AR          | H     | c.[1155dupC]+<br>c.[1155dupC]             | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56] | 45             | 156            | -1.12         | 70.7                   | 1.3                            | 19                   | 80.5                 | CKD-2                | 21  | Citrate   | Yes                 | No                   | Yes                   | No                   | Yes             | No              |  |                          | No                                  |                          |      | 18.8 |
| Male   | 0.4                        | ATP6V1B1 | AR          | CH    | c.[585+1G>A]+<br>c.[1061G>A]              | p.[?]+ p.[GLu330Lys]                      | 60.5           | 163            | -1.89         | 76                     | 0.7                            | 19                   | 78.3                 | CKD-2                | 22  | Citrate   | No                  | Yes                  | Yes                   | Yes                  | Yes             | Yes             |  | 2                        | No                                  |                          |      | 18.6 |
| Female | 0.08                       | ATP6V1B1 | AR          | H     | c.[1418T>C]+<br>c.[1418T>C]               | p.[Leu473Pro]+<br>p.[Leu473Pro]           | 64             | 151            | -1.9          | 65                     | 1.3                            | 20                   | 100.8                | CKD-1                | 25  | HCO3      | No                  | Yes                  | Yes                   | No                   | Yes             | Yes             |  | 5                        | No                                  |                          |      | 19.9 |
| Female | 1                          | ATP6V1B1 | AR          | H     | c.[1155dupC]+<br>c.[1155dupC]             | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56] | 59             | 158            | -0.82         | 49                     | 1.6                            | 21                   | 138.3                | CKD-1                | 26  | Both      | No                  | Yes                  | Yes                   | Yes                  | Yes             | Yes             |  |                          | No                                  |                          |      | 20.0 |
| Female | 5                          | ATP6V1B1 | AR          | H     | c.[585+1G>A]+<br>c.[585+1G>A]             | p.[?]+p.[?]                               | 53.5           | 156.5          | -0.98         | 63                     | 1.5                            | 22                   | 102.5                | CKD-1                | 23  | Citrate   | No                  | Yes                  | Yes                   | Yes                  | Yes             | Yes             |  | 4                        | No                                  |                          |      | 17.0 |
| Male   | 0.15                       | ATP6V1B1 | AR          | H     | c.[242T>C]+<br>c.[242T>C]                 | p.[Leu81Pro]+<br>p.[Leu81Pro]             | 97             | 172            | -0.68         | 104                    | 0.6                            | 23                   | 76.8                 | CKD-2                | 23  | Both      | No                  | Yes                  | Yes                   | Yes                  | Yes             | Yes             |  | 18                       | No                                  |                          |      | 22.9 |

| Gender | Age<br>Diagnosis<br>(year) | Gen      | Inheritance | Trait | c.DNA  | Protein result  | Weight<br>(kg) | Height<br>(cm) | Height<br>SDS | Alkali<br>serum dose<br>Creatinine (mEq/<br>μmol/L) kg/day | Age at<br>last<br>Follow Up<br>(ml/<br>min) | eGFR<br>CKD<br>Stages<br>I-V | HCO <sub>3</sub> <sup>-</sup><br>(mmol/L) | Treatment | Hyper-<br>calciuria | Metabolic<br>Control | Nephro-<br>calcinosis | Nephro-<br>lithiasis | Hearing<br>loss | Hearing<br>aids | Hearing<br>prescription<br>(age) | Cochlear<br>Implantation | Cochlear<br>prescription<br>(years) | Time<br>of FU<br>(years) |    |      |
|--------|----------------------------|----------|-------------|-------|--|---|----------------|----------------|---------------|--|---|------------------------------|---|-----------|---------------------|----------------------|-----------------------|----------------------|-----------------|-----------------|----------------------------------|--------------------------|-------------------------------------|--------------------------|----|------|
|        |                            |          |             |       |  |   |                |                |               |  |   |                              |   |           |                     |                      |                       |                      |                 |                 |                                  |                          |                                     |                          |    |      |
| Male   | 1                          | ATP6V1B1 | AR          | H     | c.[1155dupC]+<br>c.[1155dupC]<br>c.[539G>A]+                             | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56]<br>p.[Gly180Asp]+ | 65             | 174            | -0.3          | 352.9  | 1.1   | 25                           | 18.4                                      | CKD-4     | 16.8                | HCO3                 | No                    | No                   | No              | No              | Yes                              | No                       |                                     | No                       |    | 24.0 |
| Female | 2.5                        | ATP6V1B1 | AR          | CH    | c.[814G>C]<br>c.[1155dupC]+<br>c.[1155dupC]                              | p.[Ala272Pro]<br>p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56]  | 74             | 158            | -0.82         | 91   | 1.9   | 28                           | 63.8                                      | CKD-2     | 21                  | Citrate              | No                    | No                   | Yes             | Yes             | Yes                              | Yes                      |                                     | 5 No                     |    | 25.5 |
| Female | 0.08                       | ATP6V1B1 | AR          | H     | c.[1155dupC]+<br>c.[1155dupC]  | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56]                   | 69             | 152            | -1.74         | 62   | 1.4   | 29                           | 98.7                                      | CKD-1     | 21                  | Both                 | No                    | No                   | Yes             | No              | Yes                              | Yes                      |                                     | 6 No                     |    | 28.9 |
| Female | 0.08                       | ATP6V1B1 | AR          | H     | c.[1155dupC]+<br>c.[1155dupC]<br>c.[585+1G>A]+                           | p.[Ile386Hisfs*56]+<br>p.[Ile386Hisfs*56]<br>p.[?]+p.[?]    | 83             | 160            | -0.52         | 69   | 1   | 35                           | 84  | CKD-2     | 19                  | Both                 | No                    | No                   | Yes             | No              | Yes                              | Yes                      |                                     | 2 No                     |    | 34.9 |
| Male   | 1.8                        | ATP6V1B1 | AR          | H     | c.[585+1G>A]   | p.[?]+p.[?]   | 69             | 164.5          | 0.27          | 91   | 0.5   | 40                           | 80  | CKD-2     | 26.7                | HCO3                 | No                    | Yes                  | Yes             | No              | Yes                              | Yes                      |                                     | 0 No                     |    | 38.2 |
| Female | 0.2                        | N.M.I.   | No          |       |  |   | 6.7            | 65             | -1.52         | 36   | 4.8   | 0.7                          | 66  | CKD-2     | 17.6                | HCO3                 | No                    | No                   | No              | No              | No                               | No                       |                                     | No                       |    | 0.5  |
| Female | 0.5                        | N.M.I.   | No          |       | Simple het:<br>ATP6V1B1  | c.[785+1G>A]+[=]<br>p.[?]+[=]                               | 9.2            | 72             | -1.84         | 21   | 7.6   | 1.3                          | 125                                       | CKD-1     | 27                  | Both                 | No                    | Yes                  | Yes             | No              | No                               | No                       |                                     | No                       |    | 0.8  |
| Female | 1                          | N.M.I.   | No          |       |  |   | 9.6            | 81             | -0.77         | 25.6   | 2.8   | 1.8                          | 115                                       | CKD-1     | 23.8                | Citrate              | No                    | Yes                  | Yes             | No              | Yes                              | No                       |                                     | No                       |    | 0.8  |
| Female | 1                          | N.M.I.   | No          |       | Simple het:<br>ATP6V0A4  | c.[2035G>T]+[=]<br>p.[Asp679Tyr]+[=]                        | 11.5           | 89             | 0.8           | 20.3   | 5.2   | 2                            | 160                                       | CKD-1     | 22.1                | HCO3                 | No                    | Yes                  | Yes             | No              | No                               | No                       |                                     | No                       |    | 1.0  |
| Female | 1                          | N.M.I.   | No          |       | Simple het:<br>ATP6V1B1  | c.[196G>A]+[=]<br>p.[Val66Ile]+[=]                          | 12.1           | 82.1           | -3.18         | 35   | 2.6   | 3                            | 86  | CKD-2     | 24                  | Citrate              | No                    | Yes                  | Yes             | Yes             | Yes                              | Yes                      |                                     | 2 No                     |    | 2.0  |
| Male   | 0.5                        | N.M.I.   | No          |       |  |   | 11             | 82             | -3.81         | 20.5   | 4.5   | 3                            | 146                                       | CKD-1     | 20                  | Citrate              | Yes                   | No                   | Yes             | No              | No                               | No                       |                                     | No                       |    | 2.5  |
| Female | 0.5                        | N.M.I.   | No          |       |  |   | 18             | 105            | 2.66          | 31   | 2.7   | 3                            | 124                                       | CKD-1     | 23                  | Citrate              | No                    | Yes                  | Yes             | No              | No                               | No                       |                                     | No                       |    | 2.5  |
| Female | 0.1                        | N.M.I.   | No          |       | Simple het:<br>ATP6V0A4  | c.[292-1G>A]+[=]<br>p.[?]+[=]                               | 16             | 100.5          | 0.39          | 27.4   | 12.8  | 3.8                          | 134                                       | CKD-1     | 23                  | Both                 |                       | Yes                  | No              | No              | No                               | No                       |                                     | No                       |    | 3.7  |
| Male   | 0                          | N.M.I.   | No          |       | Simple het:<br>ATP6V1B1  | c.[1181G>A]+[=]<br>p.[Arg394Gln]+[=]                        | 12.3           | 95             | -1.78         | 53   | 3.3   | 4                            | 65  | CKD-2     | 19                  | Both                 | No                    | No                   | Yes             | Yes             |                                  | No                       |                                     | No                       |    | 4.0  |
| Male   | 0.1                        | N.M.I.   | No          |       | Simple het:<br>ATP6V1B1  | c.[1181G>A]+[=]<br>p.[Arg394Gln]+[=]                        | 20.8           | 106.3          | -0.1          | 26.5   | 7.2   | 4.6                          | 146                                       | CKD-1     | 27                  | Both                 | No                    | Yes                  | Yes             | No              | No                               | No                       |                                     | No                       |    | 4.5  |
| Male   | 0.16                       | N.M.I.   | No          |       |  |   | 18             | 105.5          | -0.22         | 53   | 4.1   | 4.7                          | 73  | CKD-2     | 21                  | Both                 | No                    | No                   | Yes             | No              | No                               | No                       |                                     | No                       |    | 4.5  |
| Male   | 0.25                       | N.M.I.   | AR          |       | Simple het:<br>ATP6V0A4  | c.[1185delC]+[=]<br>p.[Tyr396Thrfs*12]<br>+[=]              | 19.6           | 119.5          | -0.38         | 28   | 2.4   | 7                            | 156                                       | CKD-1     | 25                  | Both                 | Yes                   | No                   | Yes             | No              | No                               | No                       |                                     | No                       |    | 6.8  |
| Female | 1                          | N.M.I.   | AR          |       | Simple het:<br>ATP6V1B1  | c.[1394G>A]+[=]<br>p.[Arg465His]+[=]                        | 22             | 118            | -0.69         | 44.2   | 7.3   | 7                            | 97  | CKD-1     | 22                  | Both                 | Yes                   | No                   | No              | No              | No                               | No                       |                                     | No                       |    | 6.0  |
| Male   | 3                          | N.M.I.   | No          |       |  |   | 21             | 117            | -1.97         | 47.7   | 5.1   | 8                            | 90  | CKD-1     | 24                  | Citrate              | No                    | Yes                  | Yes             | No              | Yes                              | No                       |                                     | Yes                      | 4  | 5.0  |
| Male   | 1.5                        | N.M.I.   | No          |       |  |   | 27.7           | 124            | -0.72         | 43   | 4.2   | 8                            | 105                                       | CKD-1     | 20.5                | Both                 | No                    | No                   | No              | No              | No                               | No                       |                                     | No                       |    | 6.5  |
| Female | 4                          | N.M.I.   | No          |       | Distal<br>trisomy 17q<br>(mosaicism)<br>(q34.3;q21) [10]<br>/46 XX [12]) | (46, XX, der(9)t(9;17)<br>?)<br>p.[Val426Met]+p.[=]         | 14.5           | 101            | -5.54         | 34   | 1.2   | 8.5                          | 108                                       | CKD-1     | 28.8                | HCO3                 | No                    | Yes                  | Yes             | No              | Yes                              | Yes                      |                                     | 4 No                     |    | 4.5  |
| Male   | 5                          | N.M.I.   | AR          |       | Simple het:<br>ATP6V1B1  | c.[1276G>A]+[=]<br>p.[Val426Met]+p.[=]                      | 26             | 130            | -0.61         | 51.2   | 0.8   | 9                            | 93  | CKD-1     | 26                  | HCO3                 | No                    | Yes                  | Yes             | No              | Yes                              | Yes                      |                                     |                          |    | 4.0  |
| Male   | 4                          | N.M.I.   | No          |       |  |   | 24             | 121            | -2.12         | 66.3   | 3.8   | 9                            | 67  | CKD-2     | 25                  | Citrate              | Yes                   | No                   | Yes             | Yes             | Yes                              | Yes                      |                                     | 5 No                     |    | 5.0  |
| Male   | 0                          | N.M.I.   | No          |       |  |   | 28.1           | 152.5          | 2.99          | 61   | 1.6   | 9                            | 91  | CKD-1     | 25.2                | Both                 | No                    | Yes                  | Yes             | No              | Yes                              | No                       |                                     | No                       |    | 9.0  |
| Female | 1.5                        | N.M.I.   | No          |       |  |   | 35.8           | 145            | 0.99          | 49   | 2   | 10                           | 108                                       | CKD-1     | 25                  | Both                 | No                    | Yes                  | No              |                 | No                               | No                       |                                     | No                       |    | 8.5  |
| Female | 0                          | N.M.I.   | AR          |       | Simple het:<br>ATP6V1B1  | c.[340C>T]+[=]<br>p.[Arg114*]+p.[=]                         | 41             | 160            | 2.13          | 46   | 0.7   | 11                           | 127                                       | CKD-1     | 25.7                | Citrate              |                       |                      | No              | No              | No                               | No                       |                                     | No                       |    | 11.0 |
| Female | 1                          | N.M.I.   | No          |       |  |   | 36.2           | 142.1          | -0.31         | 46   | 1   | 11                           | 113                                       | CKD-1     | 25.4                | Citrate              | No                    | Yes                  | Yes             | No              | No                               | No                       |                                     | No                       |    | 10.0 |
| Female | 9                          | N.M.I.   | No          |       |  |   | 42             | 144            | -0.04         | 41.5   | 3.3   | 11                           | 127                                       | CKD-1     | 31.3                | Both                 | No                    | Yes                  | Yes             | Yes             | No                               | No                       |                                     | No                       |    | 2.0  |
| Male   | 0.08                       | N.M.I.   | No          |       |  |   | 52             | 151            | -0.29         | 54   | 0.7   | 12.6                         | 102                                       | CKD-1     | 25                  | HCO3                 | No                    | Yes                  | No              | No              | No                               | No                       |                                     | No                       |    | 12.5 |
| Female | 2                          | N.M.I.   | No          |       | Simple het:<br>ATP6V0A4  | c.[523G>A]+[=]<br>p.[Gly175Ser]+[=]                         | 33             | 142            | -2.2          | 34   | 1.1   | 13                           | 152                                       | CKD-1     | 24                  | HCO3                 | Yes                   | No                   | No              | No              |                                  | No                       |                                     | No                       |    | 11.0 |
| Male   |                            | N.M.I.   | No          |       | Simple het:<br>ATP6V1B1  | c.[1181G>A]+[=]<br>p.[Arg394Gln]+[=]                        | 39.8           | 155            | -0.18         | 51   | 2.7   | 13                           | 111                                       | CKD-1     | 22                  | Citrate              | No                    | Yes                  | Yes             | No              | No                               | No                       |                                     | No                       |    |      |
| Female | 5.5                        | N.M.I.   | No          |       |  |   | 44             | 154            | -1.22         | 66   | 1.8   | 15                           | 85  | CKD-2     | 24                  | Citrate              | No                    | Yes                  | Yes             | No              | No                               | No                       |                                     | No                       |    | 9.5  |
| Female | 2                          | N.M.I.   | No          |       |  |   | 57.7           | 151.3          | -1.69         | 43   | 1.2   | 15                           | 128                                       | CKD-1     | 23                  | Citrate              | No                    | Yes                  | Yes             | No              | Yes                              | Yes                      |                                     | 7 Yes                    | 14 | 13.0 |
| Female | 1                          | N.M.I.   | No          |       |  |   | 86.8           | 158.5          | -0.55         | 81   | 0.9   | 16                           | 71  | CKD-2     | 25                  | HCO3                 | No                    | Yes                  | Yes             | No              | Yes                              | No                       |                                     | No                       |    | 15.0 |
| Female | 2                          | N.M.I.   | No          |       | Simple het:<br>ATP6V1B1  | c.[1181G>A]+[=]<br>p.[Arg394Gln]+[=]                        | 55.5           | 167.7          | 0.76          | 61.9   | 1.3   | 17.6                         | 99  | CKD-1     | 23.9                | Both                 | No                    | Yes                  | Yes             | No              | Yes                              | Yes                      |                                     | 7.5 No                   |    | 15.6 |
| Female | 1.5                        | N.M.I.   | No          |       |  |   | 46             | 150            | -2.03         | 66   | 2.1   | 18.4                         | 83  | CKD-2     | 22.8                | Both                 | No                    | Yes                  | Yes             | No              | No                               | No                       |                                     | No                       |    | 16.9 |
| Female | 16                         | N.M.I.   | No          |       |  |   | 55             | 158            | -0.82         | 61.9   | 0.4   | 20                           | 106.6                                     | CKD-1     | 22                  | Citrate              | No                    | Yes                  | Yes             |                 | No                               | No                       |                                     | No                       |    | 4.0  |







| Gender | Age<br>Diagnosis<br>(year) | Gen      | Inheritance | Trait | c.DNA | Protein result | Weight<br>(kg) | Height<br>(cm) | Height<br>SDS | Alkali<br>serum dose<br>Creatinine (mEq/<br>μmol/L) kg/day | Age at<br>last<br>Follow (ml/<br>min) | eGFR<br>CKD<br>Stages I-V | HCO <sub>3</sub> <sup>-</sup><br>(mmol/L) | Treatment | Hyper-<br>calciuria | Metabolic<br>Control | Nephro-<br>calcinosis | Nephro-<br>lithiasis | Hearing<br>loss | Hearing<br>aids | Hearing<br>aids       |                          | Cochlear<br>Implantation |                  | Time<br>of FU<br>(years) |      |
|--------|----------------------------|----------|-------------|-------|-------|----------------|----------------|----------------|---------------|--|---------------------------------------|---------------------------|---|-----------|---------------------|----------------------|-----------------------|----------------------|-----------------|-----------------|-----------------------|--------------------------|--------------------------|------------------|--------------------------|------|
|        |                            |          |             |       |       |                |                |                |               |  |                                       |                           |   |           |                     |                      |                       |                      |                 |                 | prescription<br>(age) | Cochlear<br>Implantation | prescription<br>(years)  | of FU<br>(years) |                          |      |
| Female | 0.1                        | Untested | No          |       |       |                | 19             | 115            | 0             | 37.1   | 2.4                                   | 6                         | 113                                       | CKD-1     | 24.3                | Citrate              |                       |                      | Yes             | No              | Yes                   | Yes                      | 4                        | No               |                          | 5.9  |
| Female | 2                          | Untested | No          |       |       |                | 14.5           | 92             | -5.09         | 54.8   | 2.1                                   | 6                         | 61  | CKD-2     | 22                  | Citrate              | Yes                   | No                   | Yes             | No              | No                    | No                       |                          | No               |                          | 4.0  |
| Female | 3.5                        | Untested | No          |       |       |                | 17             | 108            | -2.27         | 48.6   | 2.8                                   | 6.7                       | 81  | CKD-2     | 19.1                | HCO3                 | No                    | No                   | Yes             | No              | No                    | No                       |                          | No               |                          | 3.2  |
| Male   | 4.1                        | Untested | No          |       |       |                | 17.3           | 107.7          | -2.26         | 35.4   | 5.2                                   | 6.7                       | 111                                       | CKD-1     | 22.5                | Citrate              | No                    | Yes                  | Yes             | No              | Yes                   | Yes                      | 4.2                      | No               |                          | 2.6  |
| Female | 7                          | Untested | No          |       |       |                | 15.3           | 108            | -2.67         | 32   | 2                                     | 7                         | 123                                       | CKD-1     | 20                  | HCO3                 | No                    | No                   | Yes             | No              |                       | No                       |                          | No               |                          | 0.0  |
| Female | 0.6                        | Untested | No          |       |       |                | 31             | 131.5          | 1.76          | 44.2   | 2.6                                   | 7                         | 109                                       | CKD-1     | 24                  | Citrate              | No                    | Yes                  | No              | No              | No                    | No                       |                          | No               |                          | 6.4  |
| Male   | 1                          | Untested | No          |       |       |                | 18.8           | 114            | -1.49         | 35.4   | 745.7                                 | 7                         | 118                                       | CKD-1     | 21.6                | Both                 | No                    | No                   | Yes             | No              | No                    | No                       |                          | No               |                          | 6.0  |
| Female | 6                          | Untested | No          |       |       |                | 14             | 90             | -6.86         | 30   | 6.4                                   | 7                         | 110                                       | CKD-1     | 22                  | Both                 | No                    | Yes                  | Yes             | No              | No                    | No                       |                          | No               |                          | 1.0  |
| Female | 5.2                        | Untested | No          |       |       |                | 17             | 105            | -3.85         | 26.5   | 4.2                                   | 7.5                       | 145                                       | CKD-1     | 20.5                | HCO3                 | No                    | No                   | Yes             | No              | No                    | No                       |                          | No               |                          | 2.3  |
| Female | 0.01                       | Untested | No          |       |       |                | 22.3           | 121.5          | -0.6          | 70.7   | 0.8                                   | 7.6                       | 63  | CKD-2     | 22                  | Citrate              | No                    | Yes                  | Yes             | No              | No                    | No                       |                          | No               |                          | 7.6  |
| Female | 0.1                        | Untested | No          |       |       |                | 28             | 121            | -1.2          | 42.4   | 2.3                                   | 8                         | 104                                       | CKD-1     | 23.3                | Citrate              | No                    | Yes                  | Yes             | No              | No                    | No                       |                          | No               |                          | 7.9  |
| Female | 0                          | Untested | No          |       |       |                | 19.5           | 118            | -1.75         | 44.2   | 3.6                                   | 8                         | 97  | CKD-1     | 21                  | Both                 | No                    | No                   | Yes             | No              | No                    | No                       |                          | No               |                          | 8.0  |
| Male   | 0                          | Untested | No          |       |       |                | 27             | 126            | -0.37         | 65   | 2.1                                   | 8                         | 71  | CKD-2     | 21.2                | Both                 | No                    | No                   | Yes             | Yes             | No                    | No                       |                          | No               |                          | 8.0  |
| Male   | 0.25                       | Untested | No          |       |       |                | 22.8           | 127            | -0.44         | 46.9   | 1.3                                   | 8.3                       | 99  | CKD-1     | 26                  | Citrate              | No                    | Yes                  | Yes             | Yes             | No                    | No                       |                          | No               |                          | 8.0  |
| Female | 4.5                        | Untested | No          |       |       |                | 21.5           | 123            | -1.29         | 62   | 2.5                                   | 8.5                       | 72  | CKD-2     | 19.5                | Citrate              | No                    | No                   | Yes             | No              | No                    | No                       |                          | No               |                          | 4.0  |
| Female | 0.5                        | Untested | No          |       |       |                | 23             | 120            | -1.82         | 46.9   | 1.6                                   | 8.5                       | 93  | CKD-1     | 25                  | Citrate              | No                    | Yes                  | Yes             | No              | No                    | No                       |                          | No               |                          | 8.0  |
| Female | 1.1                        | Untested | No          |       |       |                | 37             | 135            | 0.72          | 44   | 4.9                                   | 8.5                       | 112.0                                     | CKD-1     | 19                  | Citrate              | No                    | No                   | Yes             | No              |                       | No                       |                          | No               |                          | 7.4  |
| Male   | 3                          | Untested | No          |       |       |                | 30             | 135            | 0.2           | 48.6   | 3.6                                   | 9                         | 101                                       | CKD-1     | 22.4                | Citrate              | Yes                   | No                   | No              | No              | No                    | No                       |                          | No               |                          | 6.0  |
| Male   | 0.4                        | Untested | No          |       |       |                | 23             | 125            | -1.44         | 35.4   | 4.5                                   | 9                         | 129                                       | CKD-1     | 21.5                | Citrate              | No                    | No                   | Yes             | No              | Yes                   | Yes                      | 0.4                      | No               |                          | 8.6  |
| Male   | 8                          | Untested | No          |       |       |                | 29             | 124            | -1.61         | 22.9   | 2.8                                   | 9                         | 198                                       | CKD-1     | 23.9                | Citrate              | No                    | Yes                  | Yes             | Yes             | Yes                   | Yes                      |                          | No               |                          | 1.0  |
| Female | 0.5                        | Untested | No          |       |       |                | 17.9           | 100            | -6.03         | 30   | 6.7                                   | 9                         | 122                                       | CKD-1     | 21                  | Both                 | No                    | No                   | Yes             | No              | No                    | No                       |                          | No               |                          | 8.5  |
| Female | 0.33                       | Untested | No          |       |       |                | 29.5           | 138.5          | 0.12          | 49.5   | 2.8                                   | 10                        | 102                                       | CKD-1     | 22.9                | Citrate              | No                    | Yes                  | Yes             | No              | No                    | No                       |                          | No               |                          | 9.7  |
| Female | 0.1                        | Untested | No          |       |       |                | 53.6           | 141            | 0.41          | 48.6   | 1.7                                   | 10                        | 106                                       | CKD-1     | 24                  | Both                 |                       |                      | Yes             | No              | No                    |                          |                          | No               |                          | 9.9  |
| Male   | 0.1                        | Untested | No          |       |       |                | 50             | 153            | 2.09          | 41   | 1.2                                   | 10                        | 136                                       | CKD-1     | 20.7                | Both                 | No                    | No                   | Yes             | No              | No                    | No                       |                          | No               |                          | 9.9  |
| Male   | 0.24                       | Untested | No          |       |       |                |                |                |               | 55   |                                       | 10                        | 0   | CKD-5     | 24.2                | Both                 | No                    | Yes                  | Yes             | No              | Yes                   | Yes                      | 1                        | No               |                          | 9.8  |
| Male   | 0.4                        | Untested | No          |       |       |                | 44.8           | 140.7          | -0.16         | 56   | 2.9                                   | 10.7                      | 92  | CKD-1     | 24.6                | Citrate              | No                    | Yes                  | Yes             | No              | No                    | No                       |                          | No               |                          | 10.3 |
| Male   | 5                          | Untested | No          |       |       |                | 19.1           | 118.5          | -3.76         | 53   | 6.7                                   | 11                        | 82  | CKD-2     | 14.6                | HCO3                 | No                    | No                   | Yes             |                 | No                    | No                       |                          | No               |                          | 6.0  |
| Female | 0                          | Untested | No          |       |       |                | 30.2           | 138.7          | -0.73         | 38   | 2                                     | 11                        | 133                                       | CKD-1     | 24                  | HCO3                 | No                    | Yes                  | Yes             | No              | No                    | No                       |                          | No               |                          | 11.0 |
| Male   | 0.1                        | Untested | No          |       |       |                | 47             | 150            | 0.5           | 65   | 1.9                                   | 11.5                      | 84  | CKD-2     | 21.3                | Both                 | No                    | No                   | Yes             | No              | Yes                   | No                       |                          | No               |                          | 11.4 |
| Male   | 1                          | Untested | No          |       |       |                | 26.5           | 128.5          | -2.54         | 35.6   | 1.8                                   | 11.6                      | 132                                       | CKD-1     | 21.8                | HCO3                 | Yes                   | No                   | No              | Yes             | No                    | No                       |                          | No               |                          | 10.6 |
| Male   | 0.2                        | Untested | No          |       |       |                | 39             | 136            | -1.83         | 48.6   | 0.6                                   | 12                        | 102                                       | CKD-1     | 23.2                | HCO3                 | No                    | Yes                  | Yes             | Yes             | No                    | No                       |                          | No               |                          | 11.8 |
| Male   | 4                          | Untested | No          |       |       |                | 26             | 127            | -3.13         | 65.7   | 0.8                                   | 12                        | 71  | CKD-2     | 26                  | Citrate              | Yes                   | No                   | Yes             | Yes             | Yes                   | Yes                      | 7                        | No               |                          | 8.0  |
| Male   | 8                          | Untested | No          |       |       |                | 30             | 134.5          | -1.97         | 62   | 3.2                                   | 12                        | 79  | CKD-2     | 17                  | Citrate              | Yes                   | No                   | Yes             | No              | No                    | No                       |                          | No               |                          | 4.0  |
| Female | 11                         | Untested | No          |       |       |                | 44.5           | 149            | -0.34         | 70.7   | 1.8                                   | 12                        | 77  | CKD-2     | 19.5                | Citrate              | No                    | No                   | Yes             | Yes             | No                    | No                       |                          | No               |                          | 1.0  |
| Female | 0.58                       | Untested | No          |       |       |                | 42.2           | 140.5          | -1.89         | 53   | 0.4                                   | 12.5                      | 97  | CKD-1     | 18.7                | Citrate              | No                    | No                   | Yes             | Yes             | No                    | No                       |                          | No               |                          | 11.9 |
| Male   | 0.7                        | Untested | No          |       |       |                | 54.3           | 156            | -0.05         | 53   | 1.9                                   | 13                        | 107                                       | CKD-1     | 20.5                | Citrate              | No                    | No                   | No              | No              | No                    | No                       |                          | No               |                          | 12.3 |
| Female | 2                          | Untested | No          |       |       |                | 40             | 155            | -0.34         | 75   | 1.2                                   | 13                        | 75  | CKD-2     | 21                  | Both                 | No                    | No                   | Yes             | No              | No                    | No                       |                          | No               |                          | 11.0 |
| Male   | 0.5                        | Untested | No          |       |       |                | 30             | 148            | -1.07         | 62   | 7.1                                   | 13                        | 87  | CKD-2     | 18                  | Both                 | Yes                   | No                   | Yes             | Yes             | No                    | No                       |                          | No               |                          | 12.5 |
| Male   | 2                          | Untested | No          |       |       |                | 49.3           | 157.3          | 0.07          | 54   | 1.6                                   | 13                        | 106                                       | CKD-1     | 22                  | Both                 | No                    | Yes                  | Yes             | No              | No                    | No                       |                          | No               |                          | 11.0 |
| Male   | 0.16                       | Untested | No          |       |       |                | 56             | 156            | -0.05         | 51   | 2.1                                   | 13                        | 112                                       | CKD-1     | 25                  | Both                 | No                    | Yes                  | Yes             | No              | Yes                   | Yes                      | 5.5                      | No               |                          | 12.8 |
| Male   | 12                         | Untested | No          |       |       |                | 39.2           | 140            | -2.85         | 54.8   | 2.4                                   | 14                        | 93  | CKD-1     | 24.7                | HCO3                 | No                    | Yes                  | Yes             | No              | No                    | No                       |                          | No               |                          | 2.0  |
| Female | 0.1                        | Untested | No          |       |       |                | 79.2           | 153            | -1.14         | 79.6   | 1.5                                   | 14                        | 70  | CKD-2     | 18                  | Citrate              | No                    | No                   | Yes             | Yes             | No                    | No                       |                          | No               |                          | 13.9 |
| Female | 8                          | Untested | No          |       |       |                | 53             | 165.5          | 0.84          | 53   | 1.9                                   | 14                        | 114                                       | CKD-1     | 23                  | Citrate              | No                    | Yes                  | Yes             | No              | No                    | No                       |                          | No               |                          | 6.0  |
| Female | 3                          | Untested | No          |       |       |                | 52             | 139            | -3.27         | 44   | 2                                     | 14                        | 115                                       | CKD-1     | 22                  | Both                 | Yes                   | No                   | Yes             | Yes             | No                    | No                       |                          | No               |                          | 11.0 |
| Male   | 0                          | Untested | No          |       |       |                | 56             | 160            | -0.51         | 61.8   | 1.2                                   | 14                        | 94  | CKD-1     | 20                  | Both                 | Yes                   | No                   | Yes             | No              | Yes                   | Yes                      | 3                        | Yes              | 15                       | 14.0 |
| Male   | 1                          | Untested | No          |       |       |                | 57             | 155.8          | -1            | 61.8   | 1.1                                   | 14                        | 92  | CKD-1     | 22.6                | Both                 | No                    | Yes                  | Yes             | No              | No                    | No                       |                          | No               |                          | 13.0 |
| Female | 11                         | Untested | No          |       |       |                | 28             | 102            |               | 53   | 2.1                                   | 14                        | 70  | CKD-2     | 22.8                | Both                 | No                    | Yes                  | Yes             | Yes             | No                    | No                       |                          | No               |                          | 3.0  |
| Male   | 11                         | Untested | No          |       |       |                | 50             | 146            | -2.3          | 49.5   | 2.2                                   | 14.2                      | 108                                       | CKD-1     | 20.2                | HCO3                 | No                    | No                   | Yes             | No              | No                    | No                       |                          | No               |                          | 3.2  |
| Female | 1.5                        | Untested | No          |       |       |                | 29.5           | 140            | -3.29         | 14   | 3.3                                   | 14.5                      | 365                                       | CKD-1     | 16.7                | HCO3                 | No                    | No                   | No              | No              | No                    | No                       |                          | No               |                          | 13.0 |
| Female | 0                          | Untested | No          |       |       |                | 45             | 160            | -0.29         | 49   | 0.4                                   | 15                        | 119                                       | CKD-1     | 22                  | Citrate              |                       |                      | No              | No              | No                    | No                       |                          | No               |                          | 15.0 |
| Female | 0.1                        | Untested | No          |       |       |                | 47.2           | 150            | -1.84         | 66   | 1.5                                   | 15                        | 83  | CKD-2     | 18.6                | Both                 | No                    | No                   | Yes             | No              | No                    | No                       |                          | No               |                          | 14.9 |
| Male   | 0.11                       | Untested | No          |       |       |                | 53             | 149.5          | -2.54         | 98   | 3.1                                   | 15.3                      | 56  | CKD-3     | 25.8                | Both                 | No                    | Yes                  | Yes             | No              | No                    | No                       |                          | No               |                          | 15.2 |
| Female | 0.5                        | Untested | No          |       |       |                | 42.9           | 145            | -2.72         | 52   | 1.4                                   | 16                        | 102                                       | CKD-1     | 21.6                | HCO3                 |                       |                      | Yes             | No              | Yes                   | Yes                      | 1.5                      | Yes              | 5                        | 15.5 |
| Male   | 0.1                        | Untested | No          |       |       |                | 60             | 158            | -1.98         | 77   | 1.1                                   | 16                        | 75  | CKD-2     | 20.6                | Citrate              | Yes                   | No                   | Yes             | No              | No                    | No                       |                          | No               |                          | 15.9 |
| Male   | 1                          | Untested | No          |       |       |                | 53             | 170            | -0.48         | 82   | 1.3                                   | 16                        | 76  | CKD-2     | 20.6                | Both                 | No                    | No                   | Yes             | No              | No                    | No                       |                          | No               |                          | 15.0 |
| Female | 0.5                        | Untested | No          |       |       |                | 76             | 150            | -2.01         | 62.2   | 0.9                                   | 17.5                      | 88  | CKD-2     | 24.6                | HCO3                 | No                    | Yes                  | Yes             | Yes             | No                    | No                       |                          | No               |                          | 17.0 |
| Female | 0.8                        | Untested | No          |       |       |                | 49.1           | 169.5          | 1.06          | 79.5   | 1.5                                   | 18                        | 77.8                                      | CKD-2     | 23.9                | HCO3                 | No                    | Yes                  | Yes             | No              | Yes                   | Yes                      | 2.3                      | No               |                          | 17.2 |
| Male   | 15                         | Untested | No          |       |       |                | 65             | 174            | -0.3          | 88.8   | 2.3                                   | 18                        | 71.5                                      | CKD-2     | 21                  | Both                 |                       |                      | No              | No              |                       | No                       |                          | No               |                          | 3.0  |



## Supplemental Table 6. Treatment formulary.

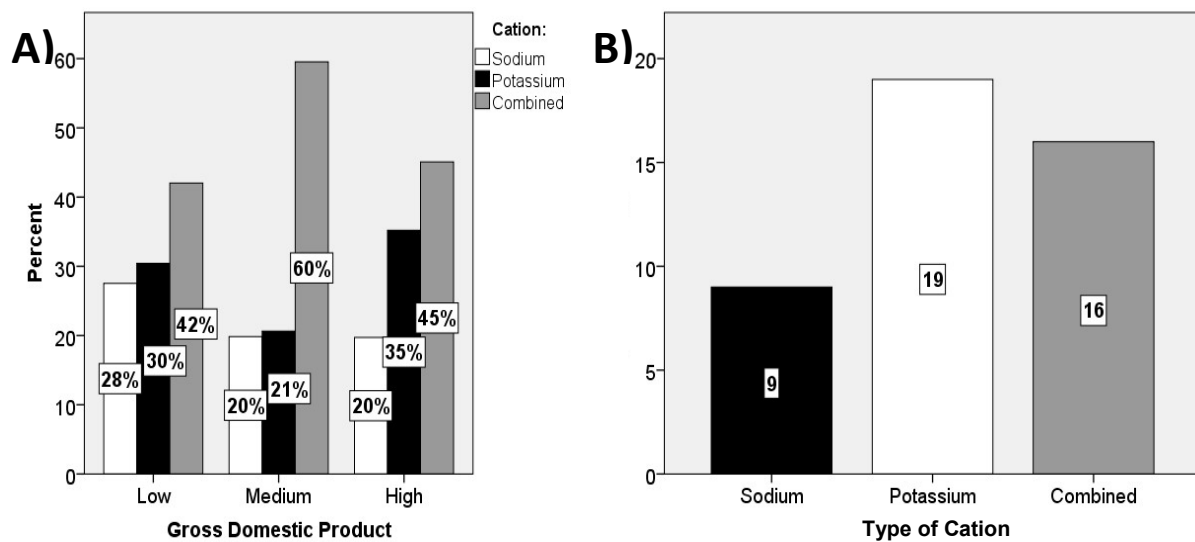
Listed are the formulations of alkali supplementation reported by the submitting clinicians and the conversion of the variously reported dosages of citrate/bicarbonate (ml, mg, sachets etc.) into molar equivalents of alkali.

| SALTS                                       | COMMERCIAL NAMES                      | FORMULATION          | DOSE   | ALKALI MMOLS   | ALKALI EQUIVALENTS |
|---|---------------------------------------|----------------------|--|--|--------------------|
| <b>Na-Citrate</b>                           | Bicitra®, Oracit®, Cytra-2®, Shohl's  | Oral Solution        | 1mL  | 0.33 mmols of Citrate  | 1 mEq              |
| <b>K-Citrate</b>                            | Cytra K®                              | Tablets / sol.       | 1100mg / 5mL                                       | 3.3 mmols of Citrate   | 10 mEq             |
|   | Cytra K®                              | Crystals             | 3300mg   | 10 mmols of Citrate  | 30 mEq             |
|   | CitraK® Forte                         | Sachets              | 2g   | 7.5 mmols of Citrate   | 22.5 mEq           |
|   | Acalka®, Urocit-K®                    | Tablets              | 1080mg   | 3.3 mmols of Citrate   | 10 mEq             |
|   | Uralyt Urate®                         | Granules             | 2.5g   | 6.6 mmols of Citrate   | 20 mEq             |
|   | Kalium-Verla®                         | Granules             | 5.4g   | 6.6 mmols of Citrate   | 20 mEq             |
|   | Urokit®                               | Sachets              | 3g   | 10 mmols of Citrate  | 30 mEq             |
|   | BioKCit®                              | Sachets              | 3.5g   | 5 mmols of Citrate   | 15 mEq             |
| BioKCit Forte®                              | Sachets                               | 4.7g                 | 6.66 mmols of Citrate                              | 20 mEq   |                    |
| <b>Na-Citrate + K-Citrate</b>               | Polycitra LC®, Tricitrates®, Cytra-3® | Oral Solution        | 5 mL<br>(550mg K-Citrate + 500mg Na-Citrate)       | 3.3 mmols of Citrate   | 10 mEq             |
|   | Uralyt-U®                             | Granules             | 2.5g   | 9 mmols of Citrate   | 27 mEq             |
|   | FONCITRIL 4000®                       | Sachets              | (1.73g K-Citrate + 1.845g Na-Citrate)              | 5.33 mmols of Citrate  | 16 mEq             |
| <b>K-Bicarbonate + K-Citrate</b>            | ADV7103® 24mEq (also 8mEq)            | Sachets              | 3660mg sachet                                      | 16 mmols of Bicarbonate + 2.61 mmols of Citrate  | 24 mEq             |
|   | Kaliner®                              | Effervescent tablets | 1 tablet (2.17g K-Citrate + 2g KHCO <sub>3</sub> ) | 6.6 mmols Citrate + 20 mmols of Bicarbonate  | 40 mEq             |
| <b>K-Bicarbonate + Na-Citrate</b>           | Blemaren® N                           | Effervescent tablets | 1 tablet   | 10 mmols of Bicarbonate + 3.3 mmols of Citrate   | 20 mEq             |
| <b>K-Citrate + Na-Bicarbonate/Carbonate</b> | Blanel®                               | Effervescent tablets | 1 tablet   | 5 mmols of Citrate + 15 mmols of HCO <sub>3</sub> <sup>-1</sup> /CO <sub>3</sub> <sup>2-</sup> | 30 mEq             |
| <b>K-Citrate + Mg-Citrate</b>               | Basica Vital E®                       | Sachets              | 5.5g   | 9.33 mmols of Citrate  | 28 mEq             |
|   | LITHOS® Prevent                       | PR Tablets           | 1 tablet (757mg K-Citrate + 358mg Mg-Citrate)      | 3.5 mmols of Citrate   | 10.5 mEq           |
|   | LITHOS® Dissolve                      | PR Tablets           | 1 tablet (1514mg K-Citrate + 581mg Mg-Citrate)     | 7 mmols of Citrate   | 21 mEq             |
|   | Lithos®                               | Sachets              | 4.5g   | 7 mmols of Citrate   | 21 mEq             |
|   | BioKMag®                              | Sachets              | 4.5g   | 7 mmols of Citrate   | 21 mEq             |
| <b>Mg-Citrate + Ca-Citrate</b>              | Cal-Mag Citrate®                      | Effervescent powder  | 5.4g (500mg Ca-Citrate + 200mg Mg-Citrate)         | 7mmols of Citrate  | 21 mEq             |
| <b>Mg-Citrate</b>                           | Magnesium Diasporal 300mg®            | Sachets              | 5g   | 8.3 mmols of Citrate   | 25 mEq             |
|   | Magnesium Diasporal 100mg®            | Tablets              |  | 2.66 mmols of Citrate  | 8 mEq              |
| <b>Mg-Citrate + K-Citrate</b>               | Lithoren®                             | Sachets              | 1 sachet   | 10 mmols of Citrate  | 30 mEq             |
| <b>Na-K-Ca-Mg Citrate</b>                   | Basica Vital®                         | Powder               | 16g  | 5.5 mmols of Citrate   | 16.5 mEq           |
| <b>Na-Bicarbonate</b>                       | Nephrotrans®                          | Tablets              | 500mg  | 6 mmols of Bicarbonate   | 6 mEq              |
| <b>K-Bicarbonate</b>                        |                                       | Tablets              | 1 g  | 10 mmols Bicarbonate   | 10 mEq             |

## Supplemental figures

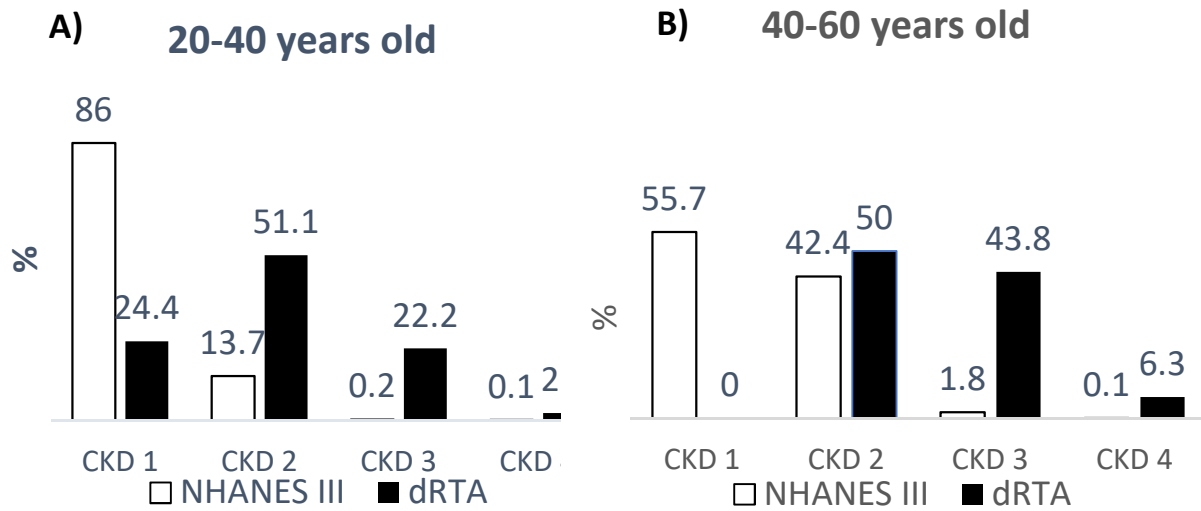
Supplemental Figure 1. Sodium versus potassium containing alkali supplementation in relation to countries GDP and presence of hypercalciuria.

**A)** Use of different type of cation in relation to gross domestic product of the countries; sodium containing salts are more frequently used in countries with low GDP, however this difference did not reach a statistical significance level. **B)** Relation between the prevalence of hypercalciuria and the type of cation used, note that patients taking sodium containing alkali did not have more hypercalciuria.



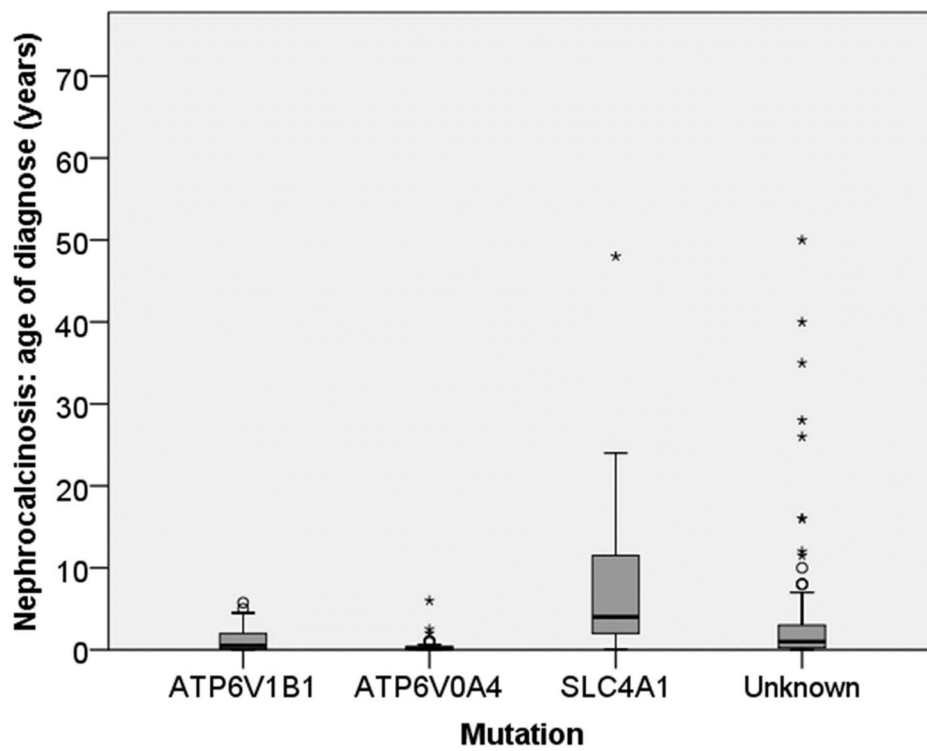
Supplemental Figure 2: CKD stratification and comparison dRTA 2017 vs NHANES III

Patients stratification according to KDIGO CKD Stages and age ranges 20-40 years (A) and 40-60 years (B). Shown is the prevalence in each CKD group for dRTA and NHANES III cohorts.



Supplemental Figure 3. Age at diagnose of nephrocalcinosis.

Detailed is the age in which nephrocalcinosis was first diagnosed. Note that virtually all patients had nephrocalcinosis when the diagnose of dRTA was made. For comparison see figure 2b (main paper).





Supplemental Figure 4: Age at presentation and per capita GDP

Age at presentation did not differ between the GDP groups. No significant difference was seen suggesting that diagnostic capabilities were similar across the GDP ranges.

