| Phenotype                                      | ICD10/UKB code | Details                                      |
|-----------------------------------------------|----------------|----------------------------------------------|
| Renal disease (ESRD or eGFR <15)              |                |                                              |
| End stage renal disease                       | N180           |                                              |
| CKD stage 5                                   | N185           |                                              |
| Complications of kidney transplant            | T861           |                                              |
| Kidney transplant status                      | Z940           |                                              |
| Dependence on renal dialysis                  | Z992           |                                              |
| Other dialysis                                | Z492           |                                              |
| Extracorporeal dialysis                       | Z491           |                                              |
| Preparatory care for dialysis                 | Z490           |                                              |
| Complications of kidney transplant            | T861           |                                              |
| Other kidney malformations                    | N27            | Small kidney                                 |
| Other congenital malformations of kidney      | Q63            |                                              |
| Malformations of ureter                       | Q621           | Congenital occlusion of ureter               |
| Congenital mega ureter                        | Q622           |                                              |
| Other obstructive defects of renal pelvis and ureter | Q623       |                                              |
| Agenesis of ureter                            | Q624           |                                              |
| Duplication of ureter                         | Q625           |                                              |
| Malposition of ureter                         | Q626           |                                              |
| Congenital vesico-uretero-renal reflux        | Q627           |                                              |
| Other congenital malformations of ureter      | Q628           |                                              |
| Kidney Cysts                                  | N281           | Cyst of kidney, acquired                     |
| Polycystic kidney, adult type                 | Q612           |                                              |
| Structural malformations of the kidney        | Q60            | Renal agenesis and other reduction defects of kidney |
| Cystic kidney disease                         | Q61            | Cystic kidney disease                        |
| Congenital hydronephrosis                     | Q620           | Congenital hydronephrosis                    |
| Lobulated, fused and horseshoe kidney         | Q631           |                                              |
| Family history of renal disease               | Z841           | Family history of disorders of kidney and ureter |
| Disorders of kidney or ureter                 | N288           | Other specified disorders of kidney and ureter |
| Disorder of kidney and ureter, unspecified    | N289           |                                              |
| eGFR | Creatinine | Cystatin C |
|------|-----------|-----------|
| ACR  | Microalbumin in urine | Creatinine (enzymatic) in urine |
| Diabetes | Glycated haemoglobin (HbA1c) | Medications - insulin, sulfonylureas, metformin |
| Years in education | Years in education |
| Fluid intelligence | Fluid intelligence |
| Income | Income |
| Job class | Job class |
| Qualifications | Qualifications |
| Urinary biomarkers | Microalbumin in urine | Creatinine (enzymatic) in urine | Potassium in urine | Sodium in urine |
| Serum biomarkers | Albumin | Alkaline phosphatase | Alanine aminotransferase | Apolipoprotein A | Apolipoprotein B | Aspartate aminotransferase | Direct bilirubin | Urea | Calcium | Cholesterol | Creatinine | C-reactive protein | Cystatin C | Gamma glutamyltransferase |
| Code | Description                        |
|------|-----------------------------------|
| 30740| Glucose                           |
| 30750| Glycated haemoglobin (HbA1c)      |
| 30760| HDL cholesterol                   |
| 30770| IGF-1                             |
| 30780| LDL direct                        |
| 30790| Lipoprotein A                     |
| 30800| Oestradiol                        |
| 30810| Phosphate                         |
| 30820| Rheumatoid factor                 |
| 30830| SHBG                              |
| 30840| Total bilirubin                   |
| 30850| Testosterone                      |
| 30860| Total protein                     |
| 30870| Triglycerides                     |
| 30880| Urate                             |
| 30890| Vitamin D                         |

**Poor mental state**

| Code | Description                        |
|------|-----------------------------------|
| 2030 | Guilty feelings                   |
| 1950 | Sensitivity/ hurt feelins         |
| 1940 | irritability                      |
| 2020 | Loneliness, isolation             |
| 1930 | Miserableness                     |
| 1920 | Mood swings                       |
| 1970 | Nervous feelings                  |
| 2040 | Risk taking                       |
| 2010 | Suffer from 'nerves'              |
| 1990 | Tense/'highly strung'             |
| 1980 | Worrier/anxious feelings          |
| 2000 | Worry too long after embarrassment|

**Intellectual disability**

| Code | Description                        |
|------|-----------------------------------|
| F70  | Mild intellectual disabilities    |
| F71  | Moderate intellectual disabilities|
| Code  | Description                                                                 |
|-------|-----------------------------------------------------------------------------|
| F72   | Severe intellectual disabilities  |
| F73   | Profound intellectual disabilities |
| F78   | Other intellectual disabilities   |
| F79   | Unspecified intellectual disabilities |
| F800  | Phonological disorder           |
| F801  | Expressive language disorder     |
| F802  | Mixed receptive-expressive language disorder   |
| F804  | Speech and language developmental delay due to hearing loss   |
| F808  | Other developmental disorders of speech and language   |
| F809  | Developmental disorders of speech and language, unspecified        |
| F810  | Specific reading disorder        |
| F812  | Mathematics disorder             |
| F818  | Other developmental disorders of scholastic skills   |
| F819  | Developmental disorders of scholastic skills, unspecified   |
| F82   | Specific developmental disorder of motor function                |

**Visited healthcare professional for psychiatric disorder**

| Code  | Description                                                                 |
|-------|-----------------------------------------------------------------------------|
| 2090  | Seen GP for nerves, anxiety, tension or depression                        |
| 2100  | Seen a psychiatrist for nerves, anxiety, tension or depression           |

**Epilepsy**

| Code  | Description                                                                 |
|-------|-----------------------------------------------------------------------------|
| 131048 | Date G40 first reported (epilepsy)                                      |
| G400  | Localisation related idiopathic epilepsy and epileptic syndromes with seizures of localised onset |
| G401  | Localisation related idiopathic epilepsy and epileptic syndromes with simple partial seizures |
| G402  | Localisation related idiopathic epilepsy and epileptic syndromes with complex partial seizures |
| G403  | Generalised idiopathic epilepsy and epileptic syndromes                    |
| G404  | Other generalised epilepsy and epileptic syndromes                          |
| G408  | Other epilepsy and recurrent seizures                                      |
| G409  | Epilepsy, unspecified                                                       |
| G40A  | Absence epileptic syndrome                                                  |
| Schizophrenia |   |   |
|---|---|---|
| F20 | Schizophrenia |
| F21 | Schizotypal disorder |
| F22 | Delusional disorders |
| F23 | Brief psychotic disorder |
| F24 | Shared psychotic disorder |
| F25 | Schizoaffective disorders |
| F28 | Other psychotic disorder not due to a substance or known physiological condition |
| F29 | Unspecified psychosis not due to a substance or known physiological condition |

| Bipolar |   |   |
|---|---|---|
| F30 | Manic episode |
| F31 | Bipolar disorder |
| F32 | Major depressive disorder, single episode |
| F33 | Major depressive disorder, recurrent |
| F34 | Persistent mood [affective] disorders |
| F38 | Other mood affective disorder |
| F39 | Unspecified mood affective disorder |
| 20122 | Bipolar disorder status |

| Congenital malformation |   |   |
|---|---|---|
| Q0 | Nervous system |
| Q1 | Eye, ear, face, neck |
| Q2 | Circulatory system |
| Q3 | Respiratory system, cleft lip and palate |
| Q4 | Digestive system |
| Q5 | Genital organs |
| Q6 | Urinary system |
| Q7 | Musculoskeletal system |
| Q8 | Other |
| Q9 | Chromosomal abnormalities, not elsewhere classified |

| Pervasive |   |   |
|---|---|---|
| F840 | Autistic disorder |
| F842 | Rett’s syndrome |
| F843 | Other childhood disintegrative disorder |
| ICD-10 Code | Description |
|-------------|-------------|
| F845        | Asperger’s syndrome |
| F848        | Other pervasive developmental disorders |
| F849        | Pervasive developmental disorder, unspecified |

**Developmental delay**

- As above: Bipolar
- As above: Schizophrenia
- As above: Pervasive
- As above: Visited healthcare professional for psychiatric disorder
- As above: Intellectual disability
- As above: Intellectual disability
- As above: Malformation
- As above: Epilepsy

**Supplementary table 1 – Phenotype definitions**
| Category                  | UKB background | 17q12 Microdeletion | 17q12 Microduplication |
|--------------------------|----------------|---------------------|------------------------|
|                          | Mean | 95%CI | n | % | Total | Mean | 95%CI | n | % | Total | Mean | 95%CI | n | % | Total |
| Participants             |      |       | 450,879 | 89.75 | 502,371 | - | - | 10 | 90.91 | 11 | - | - | 100 | 94.34 | 106 |
| Sex (male)               |      |       | 206,140 | 45.72 | 450,879 | - | - | 5 | 50 | 10 | - | - | 6 | 60 | 100 |
| Age (years)              | 57.28 | - | 57.26, 57.31 | 52.58 | 46.22-58.93 | - | - | 10 | 56.84 | 55.19-58.48 | - | - | 100 |
| BMI                      | 27.40 | 27.38-27.41 | 449,140 | 63.95 | 22.86-26.88 | - | - | 10 | 28.35 | 27.27-29.43 | - | - | 100 |
| Diabetes                 | - | - | 30,191 | 6.70 | 450,879 | - | - | 6 | 60 | 10 | - | - | 9 | 90 | 100 |
| eGFR                     | 92.72 | - | 92.67-92.87 | 50.59-77.32 | 77.66 | 73.96-81.37 | - | - | 9 | 77.66 | - | - | 4 |
| ACR                      | 1.98 | 1.95-2.01 | 437,935 | 2.32 | 0.62-4.03 | - | - | 9 | 5.41 | 1.67-9.15 | - | - | 95 |
| ALP                      | 83.59 | 83.52-83.67 | 429,706 | 63.95 | 50.59-77.32 | - | - | 9 | 77.66 | 73.96-81.37 | - | - | 94 |
| ALT                      | 23.54 | 23.43-23.68 | 429,706 | 49.24 | 12.42-86.07 | - | - | 9 | 24.79 | 22.25-27.33 | - | - | 94 |
| GGT                      | 37.37 | 37.24-37.50 | 429,706 | 112.0 | 10.33-213.72 | - | - | 9 | 40.82 | 34.78-46.86 | - | - | 94 |
| Smoking                  | - | - | 202,204 | 45.46 | 444,793 | - | - | 4 | 40 | 10 | - | - | 50 | 51.02 | 98 |
| Obesity                  | - | - | 110,326 | 24.47 | 450,879 | - | - | 0 | 0 | 10 | - | - | 30 | 30 | 100 |
| CKD Family history       | - | - | 61 | 0.01 | 450,879 | - | - | 0 | 0 | 10 | - | - | 0 | 0 | 100 |
| Hypertension             | - | - | 241,572 | 23.94-23.56 | 429,706 | - | - | 3 | 30 | 10 | - | - | 59 | 59 | 100 |
| BP medication            | - | - | 92,981 | 20.77 | 447,703 | - | - | 3 | 30 | 10 | - | - | 29 | 29 | 100 |
| Systolic BP              | 144.24 | 144.17-144.31 | 449,857 | 136.6 | 118.94-154.26 | - | - | 10 | 146.36 | 141.24-151.48 | - | - | 100 |
| Diastolic BP             | 86.35 | 86.64-86.69 | 449,857 | 88.25 | 77.83-98.37 | - | - | 10 | 87.52 | 84.69-90.35 | - | - | 100 |
| ESRD                     | - | - | 1,339 | 0.30 | 450,879 | - | - | 1 | 10 | 10 | - | - | 2 | 2 | 100 |

**Supplementary Table 2 – Cohort clinical characteristics.** UKB background = EU ancestry excluding; 17q12 microdeletions, microduplications and *HNF1B* pathogenic mutations. Age = age at recruitment to UKB study. Smoking = if participants had ever smoked. Obesity calculated from BMI. Hypertension based on blood pressure medications and blood pressure. ESRD = eGFR < 15 plus participants with renal replacement therapies; ALP = alkaline phosphatase; AST = aspartate amino transferase; ALT = alanine aminotransferase; GGT = gamma-glutamyl transferase;