**Overview of biochemical, genetic, epigenetic and transcriptomic biomarkers and derived phenotypes currently available in the NICOLA cohort**.

|  |  |
| --- | --- |
| Biomarker | Derived Variables |
| Apolipoprotein A |  |
| Apolipoprotein B |  |
| Cholesterol |  |
| Direct low-density lipoprotein |  |
| Gamma Glutamyltransferase |  |
| High-density lipoprotein-Cholesterol |  |
| Lipoprotein (a) |  |
| Triglycerides |  |
| Alkaline phosphatase |  |
| Calcium |  |
| Rheumatoid factor |  |
| Vitamin D |  |
| Oestradiol |  |
| Sex hormone-binding globulin |  |
| Testosterone |  |
| Glucose |  |
| Creatinine | eGFR equation based on serum creatinine |
| Cystatin C | eGFR equation based on serum cystatin C |
| Phosphate | eGFR combined equation based on serum creatinine and serum cystatin C |
| Total protein | Chronic Kidney Disease |
| Urate | Chronic Kidney Disease Stage |
| Urea | End-Stage Renal Disease |
| Alanine aminotransferase |  |
| Albumin |  |
| Aspartate aminotransferase |  |
| Direct Bilirubin |  |
| Gamma Glutamyltransferase |  |
| Total Bilirubin |  |
| Infinium CoreExome-24 Array | Imputation to the 1KGP3 reference panel  Imputation to the HRC reference panel  Clinically actionable variants |
|  | **Annotated and Filtered VCF files** (Batches 1 & 2; 1KGP3 & HRC Panels)**:**  VCF files with only polymorphic variants  Lists of monomorphic variants after imputation  Quality Control information of the imputation process  VCF files with polymorphic variants annotated with RS  VCF files with polymorphic variants with R2>0.3  VCF files with polymorphic variants with R2>0.3 and MAC≥5  VCF files with polymorphic variants with R2>0.3 and MAC≥5 annotated with RS and gene |
|  | **Kinships for relatedness in association analysis:**  Kinship matrix for autosomes, 1KGP3 reference panel  Kinship matrix for chrX, 1KGP3 reference panel  Kinship matrix for autosomes, HRC reference panel  Kinship matrix for chrX, HRC reference panel |
|  | **Software specific files** (Batches 1 & 2; 1KGP3 & HRC Panels)**:**  Concatenated VCF files with polymorphic variants for kinship generation  Concatenated VCF files with polymorphic variants and duplicated SNPs removed for clumping  pgen files for input in PLINK 2.00 alpha |
|  | **Individual Genome-Wide Association Summary statistics for:**  Total cholesterol  HDL-cholesterol  LDL-cholesterol  Non-HDL cholesterol  Triglycerides  Height  Body-mass index  Waist-to-hip ratio  Early age-related macular degeneration  eGFR  Serum creatinine  Chronic kidney disease  Serum urea  Subretinal drusenoid deposits (reticular pseudodrusen)  Subretinal drusenoid deposits (reticular colour)  Macular pigment: peak height  Macular pigment: peak volume  Naevi  Arterial calibre  Venular calibre  Arteriovenous ratio  Arteriolar fractal dimension  Venular fractal dimension  Arteriolar tortuosity  Venular tortuosity |
|  | **Lists of dosage information on SNPs provided for candidate SNPs and Genetic Risk Score projects:**  55 SNPs previously associated with age-related macular degeneration  64 SNPs previously associated with arsenic levels |
|  | **Genome-Wide Association Meta-Analysis Summary statistics for:**  Subretinal drusenoid deposits (reticular pseudodrusen)  Subretinal drusenoid deposits (reticular colour)  Macular pigment: peak height  Macular pigment: peak volume  Naevi  Arterial calibre  Venular calibre  Arteriovenous ratio  Arteriolar fractal dimension  Venular fractal dimension  Arteriolar tortuosity  Venular tortuosity |
|  | Beta values (β) |
|  | M values |
|  | Proportional cell counts |
|  | Epigenetic clocks |
|  | **Summary statistics for:**  Alcohol consumption  Body-mass index  Education level  eGFR  Naevi  Physical activity  Risk preference  Serum urate  Smoking  Socioeconomic status  Subretinal drusenoid deposits (reticular pseudodrusen)  Subretinal drusenoid deposits (reticular colour)  Time preference |
|  | Gene expression counts |
|  | **Summary statistics for:**  Renal phenotypes |
| Abbreviations: 1KGP3: 1000 Genomes Phase3 v5; chr: chromosome; eGFR: estimated glomerular filtration ratio; HDL: High-density lipoproteins; HRC: Haplotype Reference Consortium; LDL: Low-density lipoproteins; VCF: Variant Call Format. | |