**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 panelClinically actionable variants |
|  | **Annotated and Filtered VCF files** (Batches 1 & 2; 1KGP3 & HRC Panels)**:**VCF files with only polymorphic variantsLists of monomorphic variants after imputationQuality Control information of the imputation processVCF files with polymorphic variants annotated with RSVCF files with polymorphic variants with R2>0.3VCF files with polymorphic variants with R2>0.3 and MAC≥5VCF 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 panelKinship matrix for chrX, 1KGP3 reference panelKinship matrix for autosomes, HRC reference panelKinship matrix for chrX, HRC reference panel |
|  | **Software specific files** (Batches 1 & 2; 1KGP3 & HRC Panels)**:**Concatenated VCF files with polymorphic variants for kinship generationConcatenated VCF files with polymorphic variants and duplicated SNPs removed for clumpingpgen files for input in PLINK 2.00 alpha |
|  | **Individual Genome-Wide Association Summary statistics for:**Total cholesterol HDL-cholesterol LDL-cholesterol Non-HDL cholesterolTriglyceridesHeightBody-mass indexWaist-to-hip ratioEarly age-related macular degenerationeGFRSerum creatinineChronic kidney diseaseSerum ureaSubretinal drusenoid deposits (reticular pseudodrusen)Subretinal drusenoid deposits (reticular colour)Macular pigment: peak heightMacular pigment: peak volumeNaeviArterial 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 heightMacular pigment: peak volumeNaeviArterial 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 consumptionBody-mass indexEducation leveleGFRNaeviPhysical activityRisk preferenceSerum urateSmokingSocioeconomic statusSubretinal 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. |