



UNIVERSITÄT  
LEIPZIG

Medizinische Fakultät

LIFE-Tag: Science in LIFE Adult

# First genome-wide association study of 99 body measures derived from 3-dimensional body scans

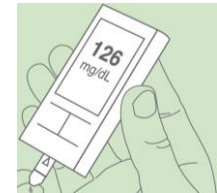
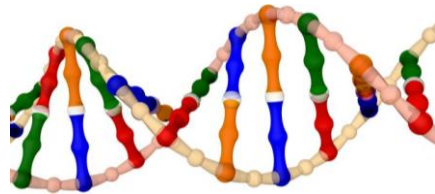
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**imise.**

# 1 MOTIVATION

- Body height, BMI, waist/hip circumference are important clinical and epidemiological risk factors/outcome variables for hypertension, T2DM, carcinoma
- Compared to body scanner (BS) traits only a few classical anthropometric (CA) traits for the characterization of the human body are available
- Additional BS traits are not investigated in more detail so far
- Molecular mechanisms behind established loci are often unclear
- Also unclear if established loci are specific for one or more (correlated) traits

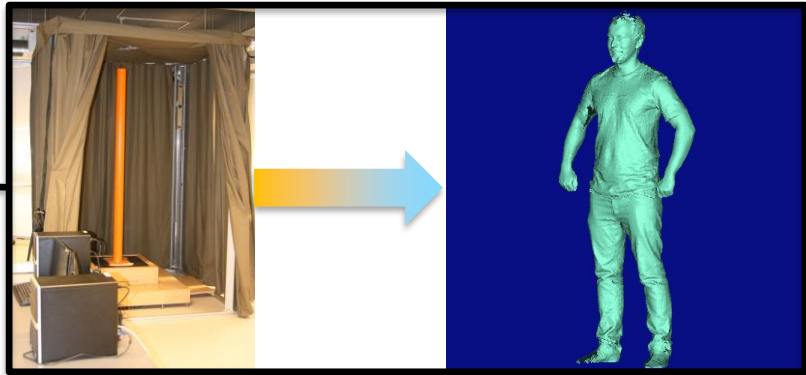


## 2 RESEARCH QUESTIONS

- I. Which anthropometric traits are affected by established genetic loci of CA traits?
- II. Are there novel loci associated with anthropometric traits?
- III. Does genetic correlation explain a common genetic origin of CA traits and BS traits?

### 3 MATERIAL: COHORT, PHENOTYPE, GENOTYPE

- LIFE-Adult: N = 10,000 individuals
- Bodyscanner VITUS XXL:
  - 155 standard measurements
  - After exclusion (reliability, to vertical, left-right): 99 traits



- Axiom Genome-Wide CEU 1 Array Plate: After QC and imputation (IMPUTE2, 1kGp3v5): N = 7,669 individuals, M = 85,063,807 SNPs

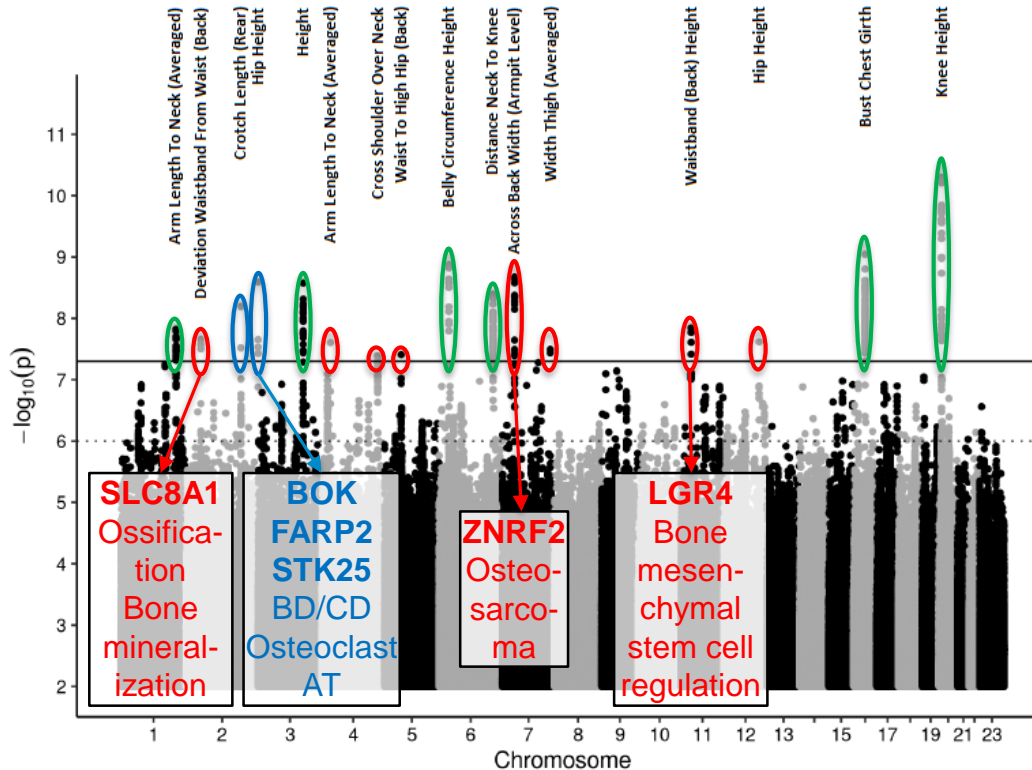
**Phenotype & Genotype data: N = 7,562 individuals from LIFE-Adult**

## 4 METHODS: STATISTICAL METHODS

1. GWAS for 99 BS traits
2. Sex-specific analysis for Top-SNPs
3. Comparison with loci of CA traits from GWAS Catalog: „*Surprise Analysis*“
4. Genetic correlation analysis

## 5 RESULTS: 1. GWAS FOR 99 BS TRAITS

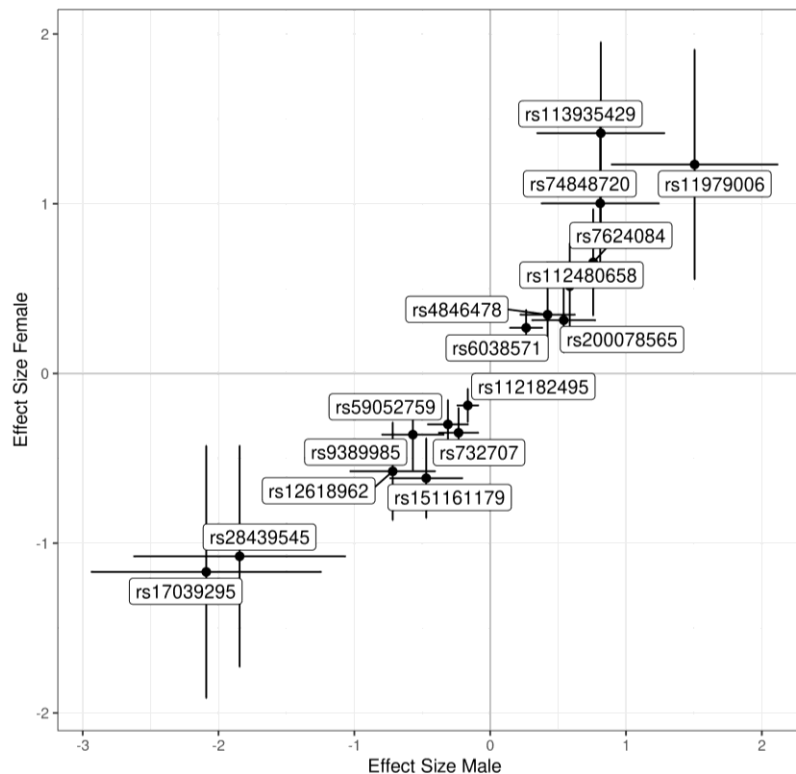
- 316 genome-wide significant associations
- distributed over 16 loci
- 45 BS traits involved
- $1.001 \leq \lambda_{GC} \leq 1.050$



- 6 loci already known for CA traits
- 2 loci already known for non-anthropometric traits
- 8 novel loci

## 5 RESULTS: 2. SEX-SPECIFIC ANALYSIS FOR TOP-SNPS

Top-SNPs of  
16 loci



No  
significant  
sex effects  
(Test for interaction)

## 5 RESULTS: 3. *SURPRISE ANALYSIS*

**Idea:** Min-Max-Correlation between CA (GWAS Catalog) and BS (GWAS)

- GWAS Catalog: 709 loci associated with CA traits ( $p \leq 5 \times 10^{-8}$ )
- GWAS: 211 of 709 loci associated with BS traits ( $p \leq p_{5\%} = 0.0011$ )

**Surprise:** Weak correlation but strong association

- **For 39 of 211 loci, the BS trait with the weakest correlation (Min-Max) was stronger associated than the BS trait with the strongest correlation.**
- **For 33 of 211 loci, the best associated BS trait was only weakly correlated, i.e.  $|r| \leq 0.3$ .**

**1p36.13**

CA: Height

BS: Distance Across Back Width (Armpit Level) To Waist ( $r=0.28$ )

**2q35**

CA: Height

BS: Hip Thigh Girth ( $r=0.25$ )

**6p24.3**

CA: Height, WHR

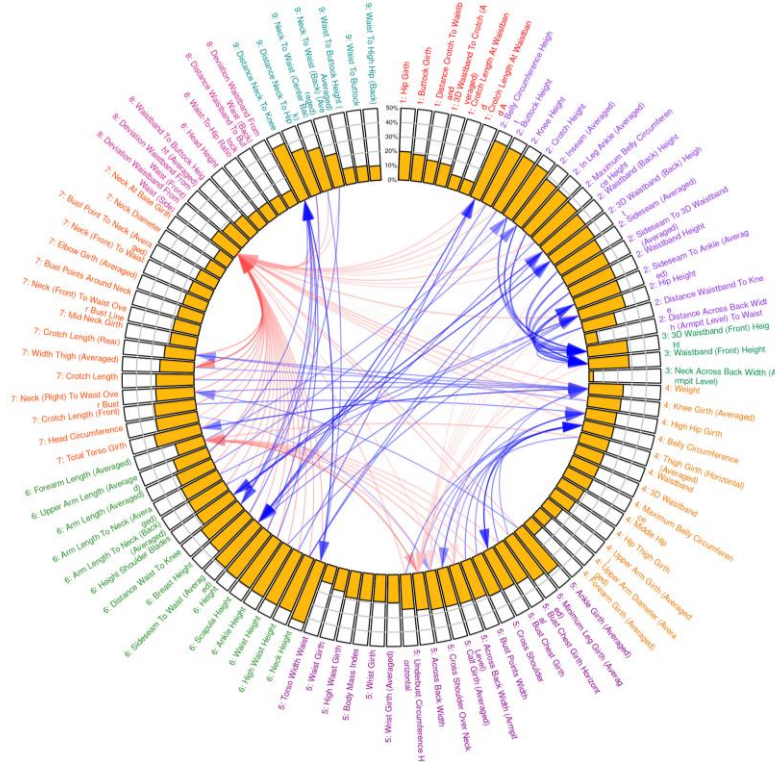
BS: Thigh Girth (Horizontal) (Averaged) ( $r=0.16, r=-0.05$ )



## 5 RESULTS: 4. GENETIC CORRELATION ANALYSIS

### Heritabilities

Body height: **39%**  
 Body weight: **24%**  
 UA length: **25%**  
 UA girth: **13%**  
 Waist girth: **15%**  
 Hip girth: **19%**  
 Thigh girth: **21%**  
 Calf girth: **27%**



### øGC in cluster

- 1 Waist/hip girths+ Distances to crotch: **29%**
- 2 Waist/hip heights+ Leg lengths: **29%**
- 3 Waistband: **23%**
- 4 Waist/hip girths+ Arm girths: **40%**
- 5 Torso girths+ Girths of extremities: **30%**
- 6 Torso lengths+ Arm lengths: **32%**
- 7 Torso girths+ Leg lengths: **43%**
- 8 Waist/hip distances: **-17%**
- 9 Torso distances: **32%**

## 6 LIMITATIONS

- Small sample size compared to published genome-wide meta-analyses of classical anthropometric traits
- No replication cohort available (body scanner not established in epidemiologic studies so far)

## 7 SUMMARY

- GWAS for 99 traits: 10 novel loci, 6 reported loci
- No sex effects for the 16 Top-SNPs
- Heritabilities: 3%-44%, 60 of 99 traits with heritability greater than 20%
- 9 cluster for 99 traits based on genetic correlation
- **Promising results for replication studies and meta-analyses (e.g. NAKO Gesundheitsstudie)**

## 8 ACKNOWLEDGEMENT

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