

# H3Africa Consortium Phenotype Harmonization WG

## Co-chairs

- Alia Benkala
- **your name here**  
(Jeff Struewing)

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# H3Africa Phenotype Harmonization

## Remit

- 1) Survey each grantee to identify phenotypes being studied at more than one site
- 2) Develop phenotyping standard (common) terms whenever possible, [REDCap]
- 3) Determine which phenotypes could be studied across the consortium (e.g., height, weight).

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## Remit (2)

4) Consider developing a “recommended” set of common phenotypes for use across the entire consortium

- ▣ “required” not appropriate (data already collected; not covered by existing consent; scientifically not justifiable; cost; etc.)

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## Remit (3)

5) Develop recommendations for which phenotypes are shared within Consortium and through European Genome-Phenome Archive (EGA)

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## Remit (4)

6) Consult with Ethics WG to develop consent language that allows data to be shared widely for other studies.

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- Cross-consortium analyses – more statistically powerful and informative
- Opportunity to make the whole greater than the sum of its part – *synergy*

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- A model to follow:

## **Phenotype harmonization and cross-study collaboration in GWAS consortia: the GENEVA experience**

**Siiri N. Bennett<sup>1,\*</sup>, Neil Caporaso<sup>2</sup>, Annette L. Fitzpatrick<sup>1,3</sup>, Arpana Agrawal<sup>4</sup>, Kathleen Barnes<sup>5</sup>, Heather A. Boyd<sup>6</sup>, Marilyn C. Cornelis<sup>7</sup>, Nadia N. Hansel<sup>5</sup>, Gerardo Heiss<sup>8</sup>, John A. Heit<sup>9</sup>, Jae Hee Kang<sup>10</sup>, Steven J. Kittner<sup>11</sup>, Peter Kraft<sup>12</sup>, William Lowe<sup>13</sup>, Mary L. Marazita<sup>14</sup>, Kristine R. Monroe<sup>15</sup>, Louis R. Pasquale<sup>10</sup>, Erin M. Ramos<sup>16</sup>, Rob M. van Dam<sup>17</sup>, Jenna Udren<sup>1</sup>, and Kayleen Williams<sup>1</sup> for the GENEVA Consortium**

*Genet Epidemiol.* 2011 April ; 35(3): 159–173. doi:10.1002/gepi.20564.

# Phenotype Harmonization Example

**TABLE II. Examples of possible (a) smoking-related questions and (b) new variables for cross-study analyses**

(a) Study (N)	Smoking-related questions	Possible responses
Study 1 (2,500)	1. Do you currently smoke cigarettes?	Y/N
	2. If yes, how many cigarettes per day?	###
Study 2 (1,200)	1. Have you smoked more than 100 cigarettes in your lifetime?	Y/N
	2. If yes, do you currently smoke?	Y/N
	3. If yes, how many packs per day do you smoke?	###
Study 3 (8,500)	1. Have you ever smoked?	Y/N
Study 4 (1,250)	1. Do you currently smoke?	Y/N
Study 5 (4,200)	1. Do you smoke?	Y/N
	2. When did you first start smoking regularly?	Past year; 1–5 years ago; >5 years ago
Study 6 (6,600)	1. Have you smoked tobacco in the past month?	Y/N
Study 7 (800)	1. Have you ever smoked regularly?	Y/N
	2. If yes, do you still smoke?	Y/N
	3. If yes, how much do you smoke a day?	1–10 cigarettes, 11–20 cigarettes, 21–30 cigarettes, > 30 cigarettes



# H3Africa Phenotype Harmonization

- What phenotypes might be studied in 2 or more studies?
  - ▣ 2 questionnaires received (recently)
    - Adu & Ojo, Rasmsay & Sankoh
    - Both are large, complex questionnaires, likely to be significant “natural” overlap [both use REDCap]
  - ▣ Being analyzed by Alia & Seydou (H3ABionet)
    - Populate a table that will grow over time

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## Examples

- ▣ Pilot questionnaires - opportunity for modification
- ▣ Baseline & Follow-up questionnaire – add module
- ▣ Questionnaires in development – opportunity to modify questions, add new ones
  
- ▣ Subjects from one study can serve as controls for another
- ▣ “Replication” cohorts
  
- ▣ Cost/benefit trade-off

# H3Africa Phenotype Harmonization

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□ Questions, Suggestions?

# H3Africa Phenotype Harmonization

## Working Group Membership

- Eyitayo Fakunle, Catherine Rossouw (Abayomi)
- Tay Croxton, Nicaise Ndembi, Alash'le Abimiku (Abimiku)
- Fatiu Arogundade, David Burke, Charlotte Osafo (Adu)
- Branwen Hennig, Corinne Merle, Aurel C. Allabi (Afollabi)
- Naomi Levitt, David Adeyemi, Ayesha Motala, Branwen Hennig (Amoah)
- Bruno Bucheton, Christianne Hertzfowler (Matovu)
- Mark Engel (Mayosi)
- James Brandful, Seydou Doumbia, Oyekanmi Nash, Yasmina Jaufeerally Fakim, Ezekiel Adebisi, Dean Everett, Alia Benkahla, Alan Christoffels (Mulder)
- Alisha Wade, Nigel Crowther (Ramsay)
- Deborah Colantuoni (NHGRI)

# H3Africa Phenotype Harmonization

Project PI	Main Phenotype	Approx Sample Size*	Questionnaire Status
Adu & Ojo	kidney disease	8,000	Finalized!
Affolabi	TB pharmacogenomics	630	nested
Amoah	Type II diabetes	24,000	Finalizing
Matovu	trypanosomiasis	2,400	
Mayosi	rheumatic heart disease	6,000	
Ramsay & Sankoh	cardiometabolic disease	12,000	Near final
Stein & Ramesar	schizophrenia	2,000	

\* Total study size - not all subjects genotyped in each study.

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