Public Response to Request For a DNA Sample for Research

Joe McPartlin, Dermot Kelleher
Dept Clinical Medicine
Institute of Molecular Medicine
Trinity College Dublin
Ireland

GeneLibrary Ireland

- *An all- Ireland control collection of blood and associated data from 10,000 participants*
- *2 jurisdictions, co-funded by R&D Office Belfast and the Health Research Board, Dublin*
- *Initiated by Molecular Medicine Ireland, Queens University Belfast and Ulster University*
- *Genomic, proteomic, metabolomic aspects of the common disease burden in Ireland*
- *Designed to harmonise internationally*

Gauging public willingness to take part in DNA research

Survey public opinion

Perceptions of Biomedical Research in Ireland
Telephone Survey 2000 people *

- **A well informed public** (*less than 10% reported they had never heard of any of the listed forms of genetic research*)
- **Confidence in the ability of *individual* doctors and nurses** (82%) **and researchers** (70%), to evaluate the risks and benefits of medical research
- **Least confidence in researchers in pharmaceutical companies** (26%)
- **Quite positive about genetic research** (70%), (‘*new genetic developments will result in cures for many diseases’*)
- **Some reservations regarding the ethics of genetic research** (42%) (‘*it was tampering with nature*’)
- **only 10% felt that researchers were motivated by selfish reasons** such as money or fame
- **52% of those who had previously taken part in medical research agreed they would take part again**
- Of those that would donate a sample, **89% preferred a linked data model** over an unlinked model

*Cousins et al(2005) *Public perceptions of biomedical research: A survey of the general population in Ireland*

,

Background to the Survey

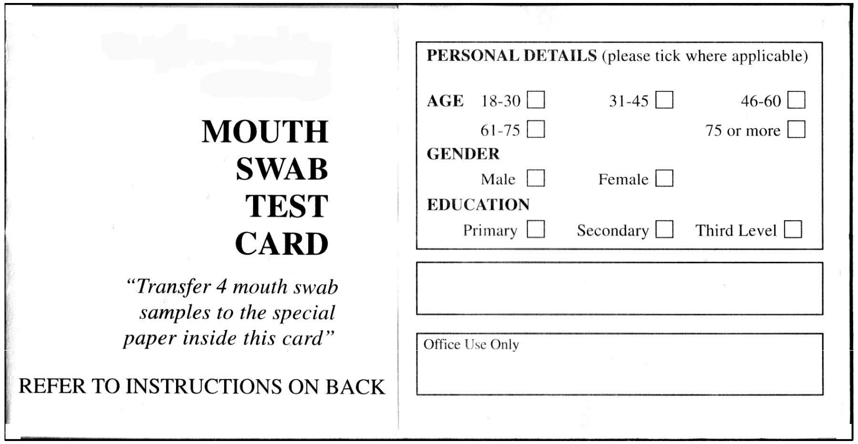
General breakdown in trust of authorities

- Health
  - *Organ Retention Scandal*
  - *Contaminated Blood Products*
- Other
  - *Property*
  - *Politicians*
  - *Priests*
  - *Police*

Testing Public Willingness

- *A well-disposed public, despite public distrust*
- *Would they be forthcoming?*
- *Would they undergo, when it was put to them, a procedure involving some effort on their part?*
- *Would they, for instance, undertake a self-administered buccal swab at home?*
- *An inexpensive undertaking but involving a degree of commitment from the public participants*

Buccal Swab Card



MOUTH
SWAB
TEST
CARD

"Transfer 4 mouth swab samples to the special paper inside this card"

REFER TO INSTRUCTIONS ON BACK

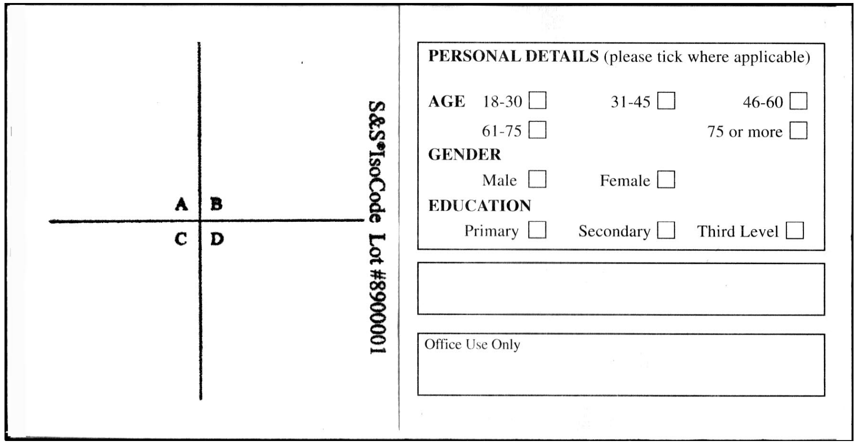
PERSONAL DETAILS (please tick where applicable)

AGE 18-30 ☐ 31-45 ☐ 46-60 ☐
61-75 ☐ 75 or more ☐

GENDER
Male ☐ Female ☐

EDUCATION
Primary ☐ Secondary ☐ Third Level ☐

Office Use Only



S&S®IsoCode Lot #8900001

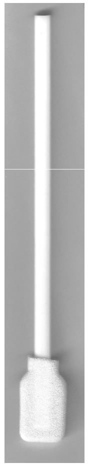
PERSONAL DETAILS (please tick where applicable)

AGE 18-30 ☐ 31-45 ☐ 46-60 ☐
61-75 ☐ 75 or more ☐

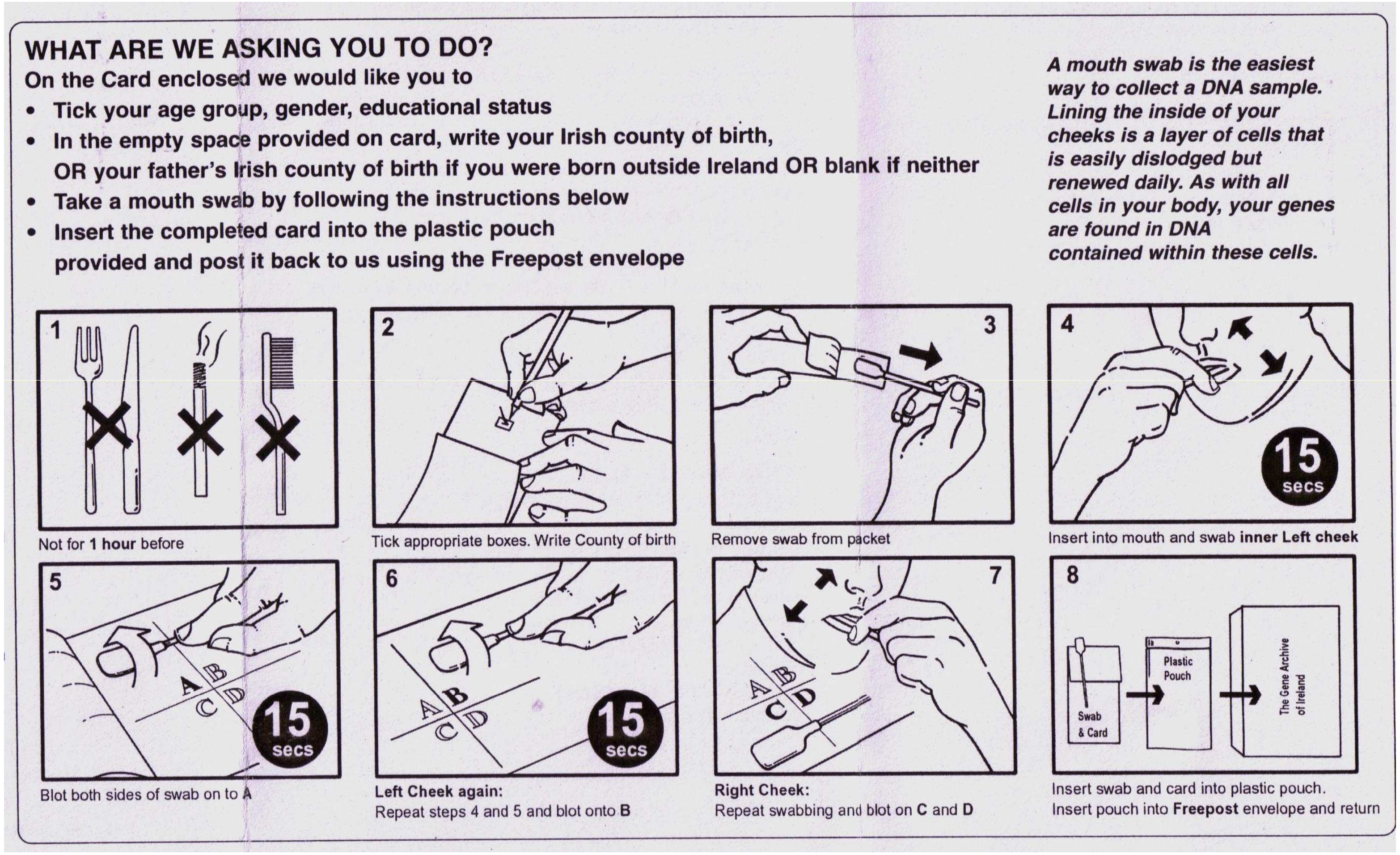
GENDER
Male ☐ Female ☐

EDUCATION
Primary ☐ Secondary ☐ Third Level ☐

Office Use Only



Instructions Cartoon*



WHAT ARE WE ASKING YOU TO DO?

On the Card enclosed we would like you to

- Tick your age group, gender, educational status
- In the empty space provided on card, write your Irish county of birth, OR your father's Irish county of birth if you were born outside Ireland OR blank if neither
- Take a mouth swab by following the instructions below
- Insert the completed card into the plastic pouch provided and post it back to us using the Freepost envelope

A mouth swab is the easiest way to collect a DNA sample. Lining the inside of your cheeks is a layer of cells that is easily dislodged but renewed daily. As with all cells in your body, your genes are found in DNA contained within these cells.

1 Not for **1 hour** before

2 Tick appropriate boxes. Write County of birth

3 Remove swab from packet

4 Insert into mouth and swab **inner Left cheek**

5 Blot both sides of swab on to **A**

6 **Left Cheek again:** Repeat steps 4 and 5 and blot onto **B**

7 **Right Cheek:** Repeat swabbing and blot on **C** and **D**

8 Insert swab and card into plastic pouch. Insert pouch into **Freepost** envelope and return

* functional illiteracy: unable to read prescription instructions on medicine (~20% population)

Requesting a DNA Sample

Focus Group: shaping the publicity literature

Group Number*	***Age***	***Sex***	***Profile***
1	18-24	M/F	B, C1, C2
2	25-45	M/F	B, C1, C2
3	46-55	M/F	B, C1, C2
4	55+	M/F	B, C1, C2

*number in each group 10-12

Ideal Hierarchy of Explanatory Messages for Participants

1. *How participants are selected*
2. *What the collection will be used for*
3. *Not involved in embryonic stem cell research or cloning*
4. *What is required to take part*
5. *Assurance of anonymity and confidentiality*
6. *Trust in organisers (who is funding the study?)*
7. *Longterm benefits to society*

Anonymised National Buccal Swab Collection

- *Buccal swab packs*
    - *Invitation letter*
    - *Buccal swab Pack*
    - *Brochure/instructions*
    - *Freepost Return Envelope*
- *Distributed by post by ESRI Random Sampling Unit*
- *Returned anonymously by participant to Trinity Biobank*
- *Repeat mail-out three weeks later*

Approved by REC Tallaght/St James Hospital,
and the Data Protection Office, Dublin

Geographical Response Proportional to Population
(r = .76)

Age Group Response

Age group (years)	***Population (1000)***	***Response (15.9%)***	***% Response***	***Response/ population***
18-30	846	175	14.6	**0.54**
31-45	961	397	33.2	**1.08**
46-60	730	347	29.0	**1.25**
61-75	424	199	16.6	**1.22**
76-	184	78	6.5	**1.12**

Gender Response

Gender	***Female***	***Male***	***Total***
N	**655**	**516**	**1171 (15.6%)**
%	**55.9**	**44.1**	

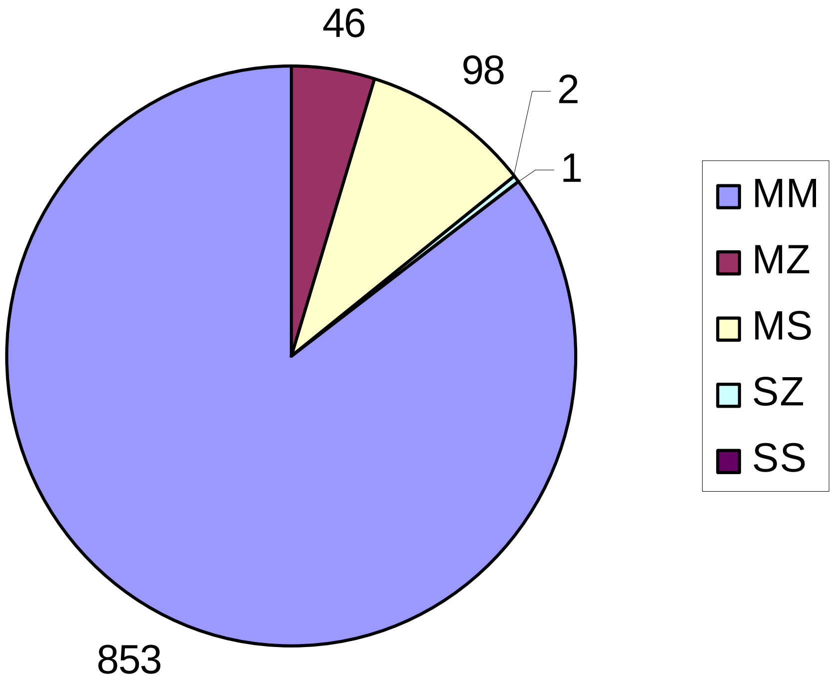
Educational-attainment Group Response

Educational Attainment	***Population (1000)***	***Response (15.6%)***	***% Response***	***Response /population***
Primary	458.1	163	13.97	**0.84**
Secondary	1577.7	458	39.20	**0.68**
Tertiary	713.9	546	46.78	**1.80**

Utility of Collected DNA

Alpha-1 Antitrypsin Deficiency: genotype frequency in the Irish Population (Carroll, T et al 2008)

- *Alpha-1 antitrypsin deficiency is caused by mutations within the AAT gene*
- *AATD is the only known genetic risk factor for the development of Chronic Obstructive Pulmonary Disease*
- ***AATD may be twice as prevalent as previously estimated, with one of the highest incidences in Europe***



-1,000 normal controls screened from TCD Buccal Swab Collection
- 46 MZ, 98 MS, 2 SZ, and 1 SS individual identified
 -Yielded high gene frequencies for S (0.053) and Z (0.022)

Buccal Swab Collection

- *Response rate 16.9%.(1267/7500)*
- *DNA quality suitable for molecular studies*
- *Uniform geographic response distribution*
- *Fewer males than females responded.*
- *The age group 18-30y responded the least, the greatest response being in the 46-60y age group.*
- *The least responsive educational group were those who attained secondary level education.*

Buccal Swab Collection

- *Though it did not involve a blood sample or the effort of attending an assessment centre, it did require the arduous task of reading the material, carrying out the test alone, and posting the sample back.*
- *With better funding, a greater uptake would have been achieved?*
- *A guide to targeting select populations for future, more comprehensive collections of biological material and associated data*