**INTRODUCTION**

- Non-communicable diseases (NCDs) are considered major public health concerns worldwide. According to the World Health Organization (WHO), NCDs are responsible for the 60 percent of total deaths globally while consequently contributing to an estimated 40 percent of universal disease burden annually (WHO Global Status Report on NCDs, 2010).

- The rise of risk factors for chronic degenerative diseases has been linked to diet, physical activity, environment, lifestyle, and genetics thus, requiring a holistic approach in order to be thoroughly studied.

- This study aims to devise a Next-Generation Sequencing (NGS) panel that will allow the sequencing of relevant genes associated with diet and nutrition-related diseases as well as to determine the allelic frequency of selected genes in the Filipino population using the NGS panel.

**MATERIALS AND METHODS**

1. **Blood collection, processing and transport**
   - Out of 10 ml of blood samples collected by trained and licensed Medical Technologists during the 8th NNS in the National Capital Region, 1.5 mL was separated and transported on ice for genomic analyses.

2. **Genomic DNA extraction**
   - A total of 187 anonymized human blood samples were used for DNA extraction.
   - Genomic DNA was isolated using the QIAamp® DNA Blood Mini Kit (Qiagen, Germany) following an optimized protocol.

3. **Targeted Sequencing**
   - Extensive Literature Review
   - Creation of Next Generation Sequencing Panel
   - Sequencing and Bioinformatics
   - Validation and Database Generation

Figure 1. The holistic approach to non-communicable diseases (NCDs) is about the integration of information obtained from different sources: environmental, clinical, biological, and genetic data.
The FNRI-designed Next-Generation Sequencing (NGS) panel examines 502 genes related to non-communicable diseases and micronutrient deficiencies.

Type 2 Diabetes Mellitus
- KCNJ11
- FABP2
- GNB3
- TNMD
- WFS3
- TCF7L2
- IL6
- SLC30A8
- DRD2

Genes
- rs5219
- rs5443
- rs10010131
- rs1800795
- rs1800497

Obesity
- APOA5
- KCTD15
- APOC3
- CETF
- APOA2
- ALPL
- ABCB1
- ADD1
- CACNA1C
- BDKR1

Genetic Variations
- rs662799
- rs2854117
- rs6265
- rs6234

Cardiovascular Diseases
- FABP2
- TNMD
- WFS3
- TCF7L2
- IL6
- SLC30A8
- DRD2

Genes
- rs5219
- rs5443
- rs10010131
- rs1800795
- rs1800497

Folate Deficiency
- MTHFR
- MAT1A
- DHFR
- SYT9

Genetic Variations
- rs1801131
- rs10887710
- rs1045642
- rs1051375

Iron Deficiency Anemia
- TMPRSS6
- AHSN
- CUBN

Genetic Variations
- rs1801131
- rs10887710
- rs1051375

**RESULTS**

Table 1. The genetic risk score of markers for folate deficiency found in the Filipino population

<table>
<thead>
<tr>
<th>Gene Symbol</th>
<th>Chr</th>
<th>snp/ID</th>
<th>Loc</th>
<th>Allele</th>
<th>Allele Count</th>
<th>MAF*</th>
<th>EAS_MAF**</th>
<th>SAS_MAF***</th>
<th>Allele Frequ. (NNS,2013)</th>
</tr>
</thead>
<tbody>
<tr>
<td>MTHFR</td>
<td>1:11854476-11854476</td>
<td>rs1801131</td>
<td>exon</td>
<td>G</td>
<td>31</td>
<td>0.2484</td>
<td>0.2192</td>
<td>0.4172</td>
<td>0.3922</td>
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<tr>
<td>MAT1A</td>
<td>11:6602797-6602797</td>
<td>rs10887710</td>
<td>intron</td>
<td>C</td>
<td>14</td>
<td>0.251</td>
<td>0.4681</td>
<td>0.3065</td>
<td>0.2353</td>
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<tr>
<td>DHFR</td>
<td>6:79934936-79934936</td>
<td>rs1643639</td>
<td>intron</td>
<td>C</td>
<td>3</td>
<td>0.2276</td>
<td>0.0466</td>
<td>0.3344</td>
<td>0.0294</td>
</tr>
<tr>
<td>SYT9</td>
<td>11:7357121-7357121</td>
<td>rs1552462</td>
<td>intron</td>
<td>T</td>
<td>2</td>
<td>0.0397</td>
<td>0.1091</td>
<td>0.0164</td>
<td>0.0392</td>
</tr>
</tbody>
</table>

*Mutation Allele Frequency; **East-Asian population; ***South-Asian population; NNS,2013 - Allele Frequency obtained from the Filipino study participants (n=187)

- The allele frequency of Methylenetetrahydrofolate Reductase (MTHFR) rs18001131 polymorphism for the Filipino population was higher than what was reported in the global frequency
- The risk variant for MTHFR rs18001131 is known to pose impaired folate metabolism.

**IN BOX: What is allele frequency?**

- Allele frequency is the relative frequency of a gene variant at a particular segment of the population (expressed as fraction or percentage). It is the fraction (or percentage) of all the chromosomes in the population that carry a given genetic variation.

Reference:

**Figure 2.** Some of the genes and their corresponding genetic variations that were enriched for targeted next-generation sequencing. The selection of genes and gene variants (n=502) was based on the published sequence-predicted functional relevance, relative to disease conditions.

**Figure 3.** The proportion of genetic markers in the FNRI-designed NGS panel with the capacity to convey meaningful information relative to frequency and diversity.
CONCLUSION & RECOMMENDATION

- A framework depicting risk scores and allelic profiles of genes and genetic variants can be generated using a Next-Generation Sequencing (NGS) panel. This panel identified the genetic biomarkers that are known to modulate the relationship between diet and health outcomes, including its incidence in the Filipino population.

- Adopting the use of the NGS panel designed by DOST-FNRI will enhance the understanding of the underlying causes of the debilitating yet preventable diseases such as Type 2 diabetes, obesity, cardiovascular disease, and other micronutrient deficiencies in the Filipino population from the genomic standpoint.

ACKNOWLEDGMENT

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