



# Literacy transition after a lecture in a pilot study of genetic result returning for the general population



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Among individuals with genetic disorders who undergo genetic analysis, it is necessary to share the results with individuals for effective prevention and treatment. However, few studies have been conducted on sharing the results of genetic analysis in the general population with limited findings regarding this practice. Therefore, the present pilot study aimed to investigate the sharing of results of genetic analysis for familial hypercholesterolemia (FH) and to investigate the rate of literacy transition after a lecture on basic genetics.

## Materials and Methods

Initially, 210 of 1,220 individuals who participated a cohort of a previous study involving whole-genome sequencing, were selected per the following inclusion criteria: adults (age > 20 years), hypercholesterolemia (total cholesterol > 250 mg/dl; LDL cholesterol > 180 mg/dl), having a past medical history, and wishing for results of genetic analysis to be shared with them. Finally, 36 out of 210 individuals consented to participate in the study and 29 valid responses were hence the final subjects (Table.1). We administered questionnaires before and after a lecture on basic genetics (Regarding DNA, chromosome, mode of inheritance etc.) and assessed their understanding using a McNemar’s test and a Wilcoxon signed rank test.



Fig. 1 Views of the genetic lecture

## Results

Table 1 Participants of familial hypercholesterolemia pilot study

|       | Candidates (n) | Documents returned (n) | Applicants (n) | Lecture attendees (n) | Study participants (n) | Participants have positive gene variants (n) | Returning the genetic results (n) |
|-------|----------------|------------------------|----------------|-----------------------|------------------------|----------------------------------------------|-----------------------------------|
| ToMMo | 175            | 96                     | 31             | 28                    | 28                     | 3                                            | 28                                |
| IMM   | 35             | 18                     | 9              | 8                     | 8                      | 4                                            | 8                                 |
| Total | 210            | 114                    | 40             | 36                    | 36                     | 7                                            | 36                                |

Table 2 Comparison of participants’ genetic knowledge before and after the genetics workshop (n=29)

|                                    | Questions                                                                              | Percentage of correct answers (%) |       |                          |
|------------------------------------|----------------------------------------------------------------------------------------|-----------------------------------|-------|--------------------------|
|                                    |                                                                                        | Before                            | After |                          |
| Q1.                                | One can see a gene with the naked eye.                                                 | 76                                | 90    | p = 0.125 <sup>1)</sup>  |
| Q2.                                | A gene is a disease.                                                                   | 35                                | 48    | p = 0.424 <sup>1)</sup>  |
| Q3.                                | A gene is a molecule that controls hereditary characteristics.                         | 38                                | 48    | p = 0.453 <sup>1)</sup>  |
| Q4.                                | Genes are inside cells.                                                                | 59                                | 86    | p = 0.021 <sup>*1)</sup> |
| Q5.                                | A gene is a piece of DNA.                                                              | 76                                | 90    | p = 0.219 <sup>1)</sup>  |
| Q6.                                | A gene is a cell.                                                                      | 24                                | 55    | p = 0.012 <sup>*1)</sup> |
| Q7.                                | A gene is a part of a chromosome.                                                      | 55                                | 79    | p = 0.065 <sup>1)</sup>  |
| Q8.                                | Different body parts include different genes.                                          | 24                                | 62    | p = 0.003 <sup>*1)</sup> |
| Q9.                                | Genes are bigger than chromosomes.                                                     | 14                                | 45    | p = 0.022 <sup>*1)</sup> |
| Q10.                               | It has been estimated that a person has 22 000 genes.                                  | 3                                 | 90    | p < 0.001 <sup>*1)</sup> |
| Q11.                               | Healthy parents can have a child with a hereditary disease.                            | 52                                | 90    | p = 0.001 <sup>*1)</sup> |
| Q12.                               | The onset of certain diseases is due to genes, environment, and lifestyle.             | 62                                | 86    | p = 0.039 <sup>*1)</sup> |
| Q13.                               | All serious diseases are hereditary.                                                   | 45                                | 79    | p = 0.013 <sup>*1)</sup> |
| Q14.                               | The child of a disease gene carrier is always also a carrier of the same disease gene. | 28                                | 66    | p = 0.013 <sup>*1)</sup> |
| First quartile of the total scores |                                                                                        | 3.0                               | 9.0   | p < 0.001 <sup>*2)</sup> |
| Third quartile of the total scores |                                                                                        | 8.5                               | 12.0  |                          |
| Median of the total scores         |                                                                                        | 6.0                               | 11.0  |                          |

1) McNemar’s test, 2) Wilcoxon signed rank test  
The questionnaire is developed based on Jallinoja & Aro (1999)

The total scores of correct answers to questions regarding basic genetics were significantly higher after than before the lecture (p < 0.001) (Table 2).

## Conclusion

The present results suggest that genetic education is effective and important for accurate understanding of genetic disorders.