Clinical implementation of Clinician-oriented locus specific mutation detection and deposition system

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Objectives

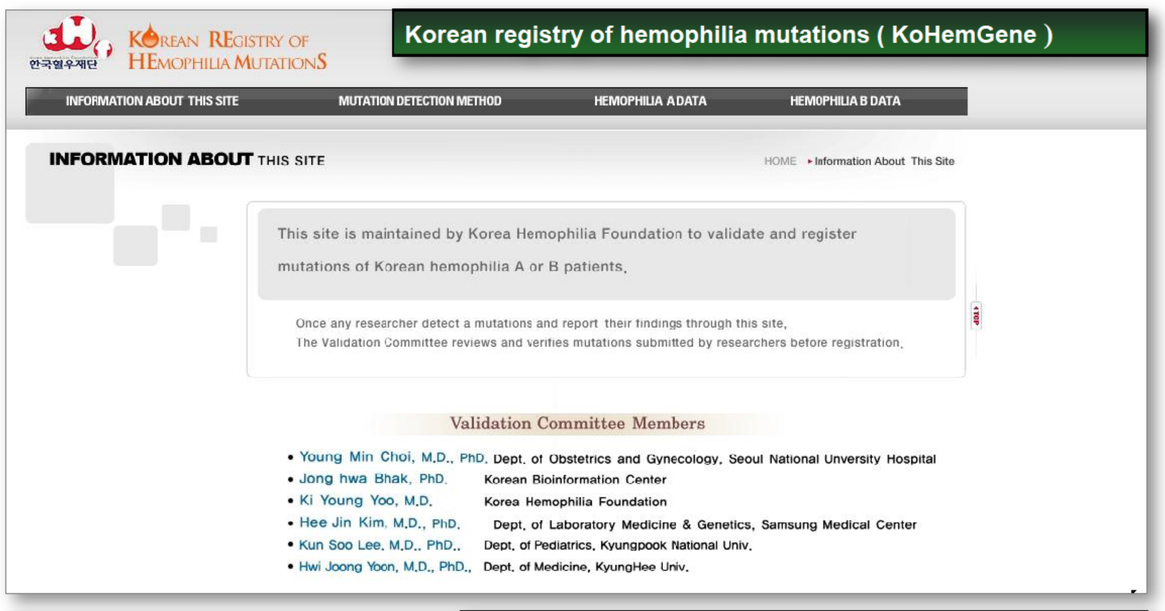
F8 gene contains 26 exons, 186kb long. Not only seauencing but also interpretation is time-consumeing and labor intensive.
So we developed the automated mutation detection software of DNA sequence, called Kohemgene(COMUS: Clinician-oriented locus specific mutation detection and deposition system). This soft ware uses UCSC genome browser as reference and is loaded in The Korea Hemophilia mutation database (www.kohemgeme.org).
This study is designed to compare KOHEMGENE and Sequencher® in terms of time-saving and accuracy of interpretation.

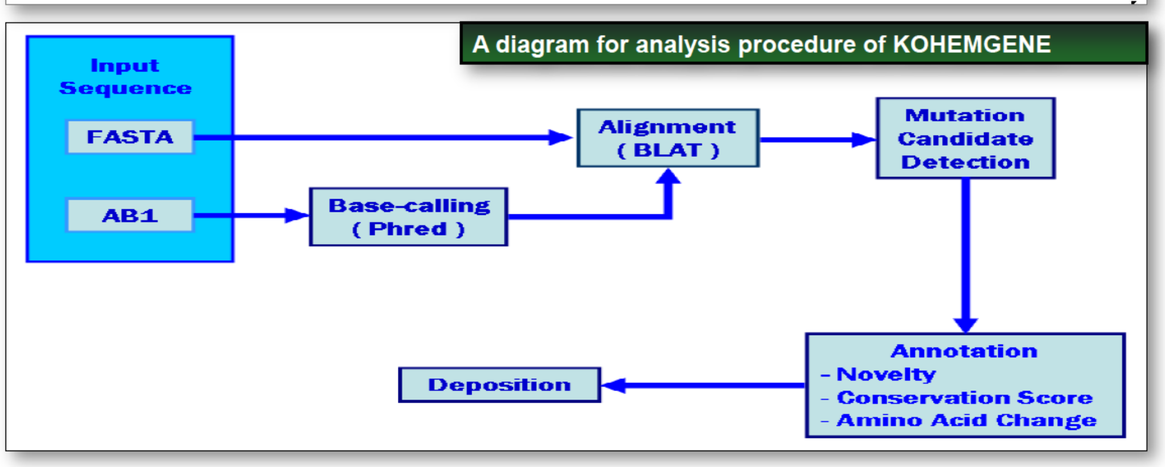
Material and Methods

We analyzed with desktop computer (Pentium® IV, 512MB) 59 unrelated hemophilia A patients not having intron 22 inversion mutation.The time for the loading of data into software to completion of interpretation was recorded in seconds. Kohengene and Sequencher® were used for the sample patient.
Meanwhile process of Sequencher® is composed 5 steps;
1)Input Ref.Sequence,
2)Input Data file,
3)Trimming,
4)data analysis
5)Report

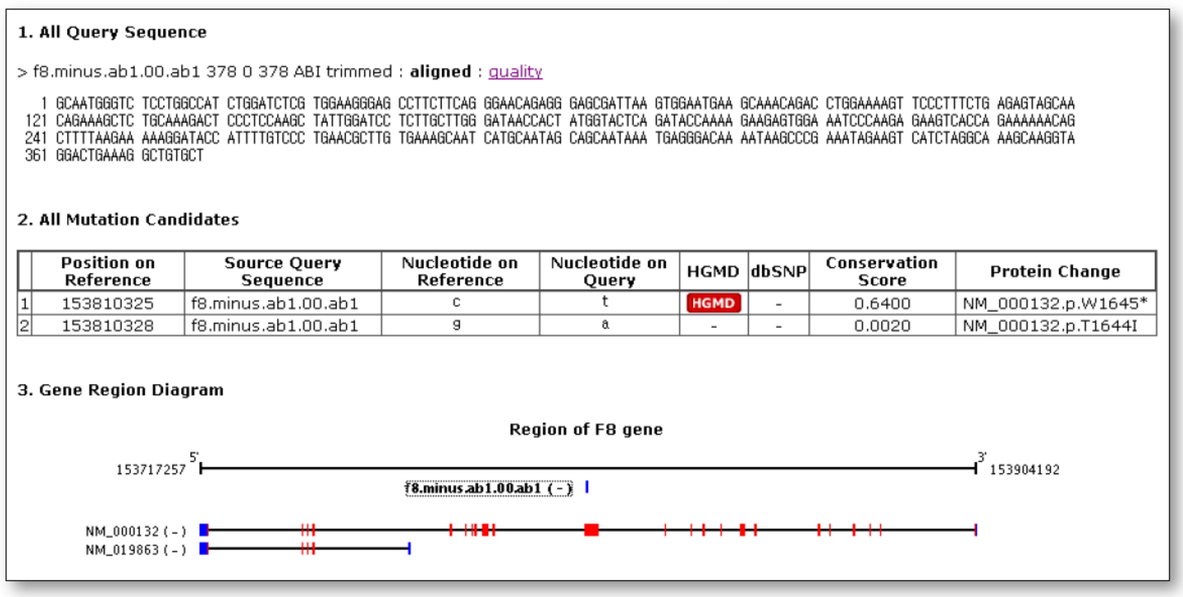
Process of Kohemgene program,
1) Input Data file (AB1 or FASTA),
2) data analysis,
3)Report.

Process of Sequencher®, Two researchers participated in the analysis, who experienced more than five years as their professional.

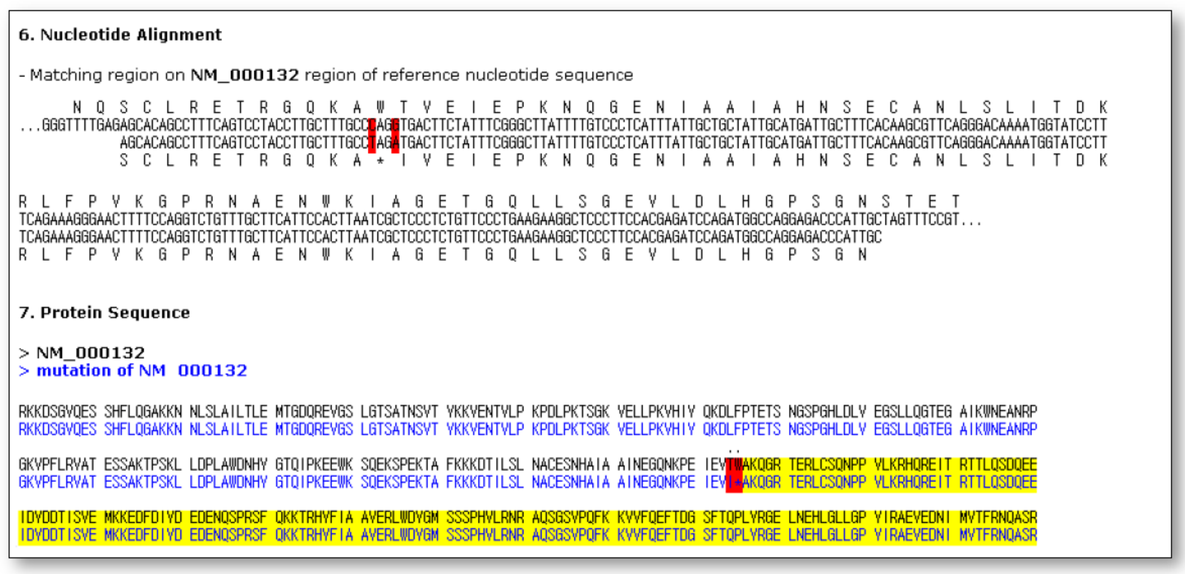




Report from KOHEMGENE



In the gene region diagram, the top line represents a gene locus of a target gene, and the blue bar below the top line represents a mapped query sequence. The next two lines represent mRNAs in the target gene locus. The blue box is UTR regions, and the red box is coding regions.



In the nucleotide alignment part, red boxes represent mutations. In the protein sequence part, red box represents mutation and yellow region represents untranslated protein sequences due to nonsense mutation.

RESULT

The result with 59 participants was matched between two programs and analysis time is reduced to 1/30 using the KOHEMGENE program.

There is no difference of interpretation between the two programs. Analysis time by using the KOHEMGENE program was 56 seconds on average. And the analysis time with Sequencher® program was 30 minutes on average.

Conclusions

The KOHEMGENE program(COMUS) provides a DNA sequence mutation analysis that helps to identify the hemophilia mutation. Also, the KOHEMGENE program(COMUS) functions automatically to search known mutation or unknown mutation in HGMD® & HAMSTeRS (Haemophilia A Mutation, Search, Test and Resource Site).

Then, researchers do not need the procedure by its help. Conclusively, the KOHEMGEME program(COMUS) facilitates research in the field of Hemophilia mutation analysis.

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