

Description

Mutations leading to expansion of GAA repeats within the first intron of the frataxin (FXN) gene are the underlying causes of Friedreich's Ataxia (FA), an autosomal recessive disorder.

The repeat numbers in normal individuals does not exceed 37, while affected individuals have higher number of GAA repeats ranging from 44 to 1700.

GT FA Detector v2 is a fluorescent-based PCR kit that detects GAA repeats in FA affected and carrier individuals via amplifying the above-mentioned triplet repeats (table 1).

Kit components

Box-A	Box-B
GT FA Detector PCR Mix	GT 1200 Size Standard
GT FA Detector PrimerMix	GTM5 Matrix Standard
GT Extend Long DNA Polymerase	
GT QCDDM Control DNA	
GT QCDFAC Carrier Control DNA	
Enhancer	
GT QCW (H2O)	

Results Interpretation

Method 1- Analyzing PCR products on 1-1.5% agarose gel along with a DNA ladder (figure 1).

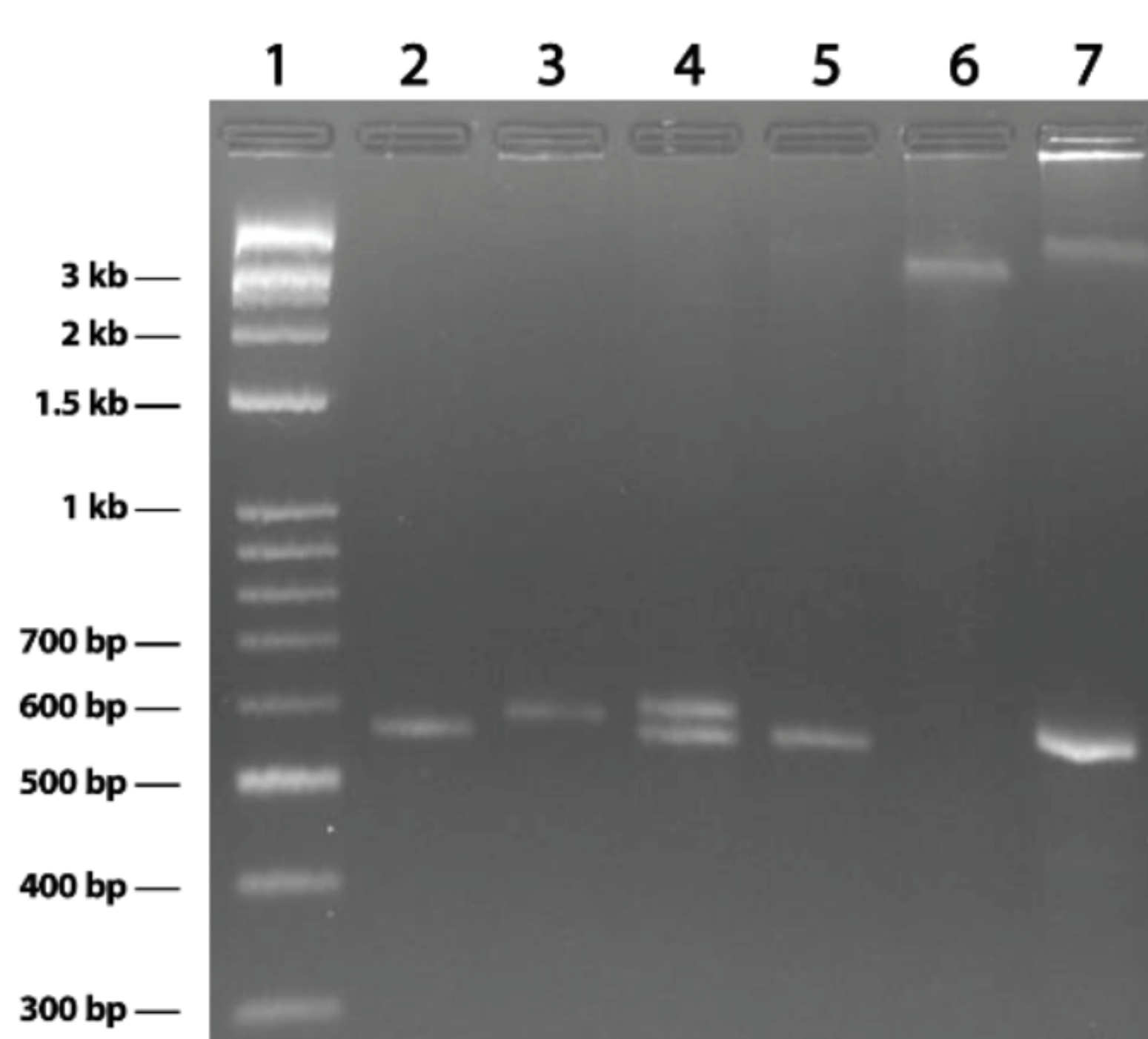


Figure 1. An Example of band patterns observed on agarose gel for FA's different genotypes amplified using **GT FA Detector v2**. Lane1: DNA ladder, Lanes2-5: normal samples, 6: affected & 7: carrier individual. As can be seen in lane 7, the mutant allele is more than 4000 bp.

Table1. GT FA Detector v2's interpretation guide.

Size	*X-543/3	GAA Repeat No.	Detectable by CE	Phenotype
558	558-543/3	5	✓	Normal allele
642	642-543/3	33	✓	
645	645-543/3	34	✓	
738	738-543/3	65	✓	Mutable normal
741	741-543/3	66	✓	
1200	1200-543/3	219	✓	Full penetrance
2343	2343-543/3	600	No	
2943	2943-543/3	800	No	
3543	3543-543/3	1000	No	
4443	4443-543/3	1300	No	

*Repeat units = Size of PCR product (bp) - 543

Method 2- More accurate repeat number determination is possible through **capillary electrophoresis analysis** using **GT1200 Size Standard** (figure 2).

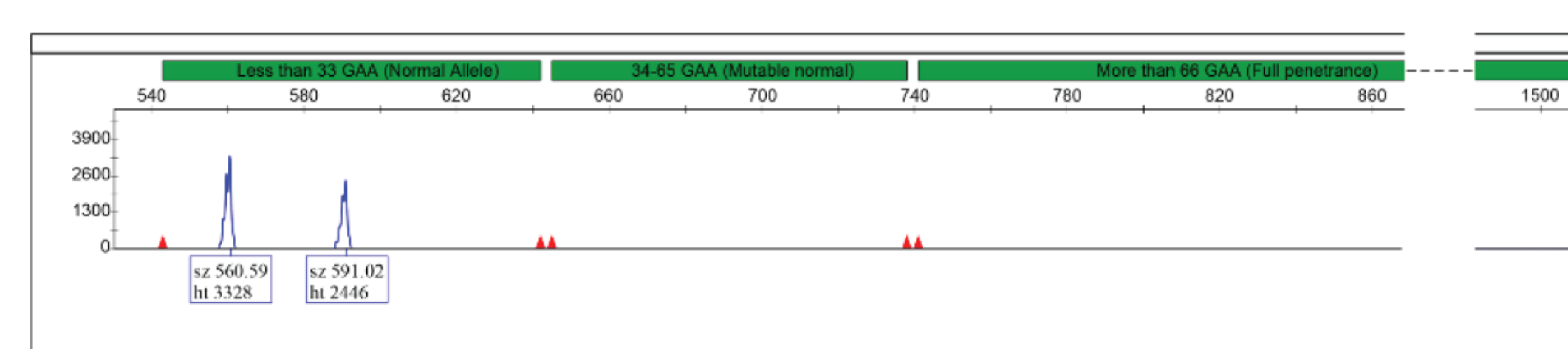


Figure 2. An Example of **GT FA Detector v2** results analyzed through capillary electrophoresis. The sample is indicative of a normal individual with 6 and 16 repeats. On an agarose gel, such heterozygote individuals are identified by their double band pattern (figure 1, lane 4)

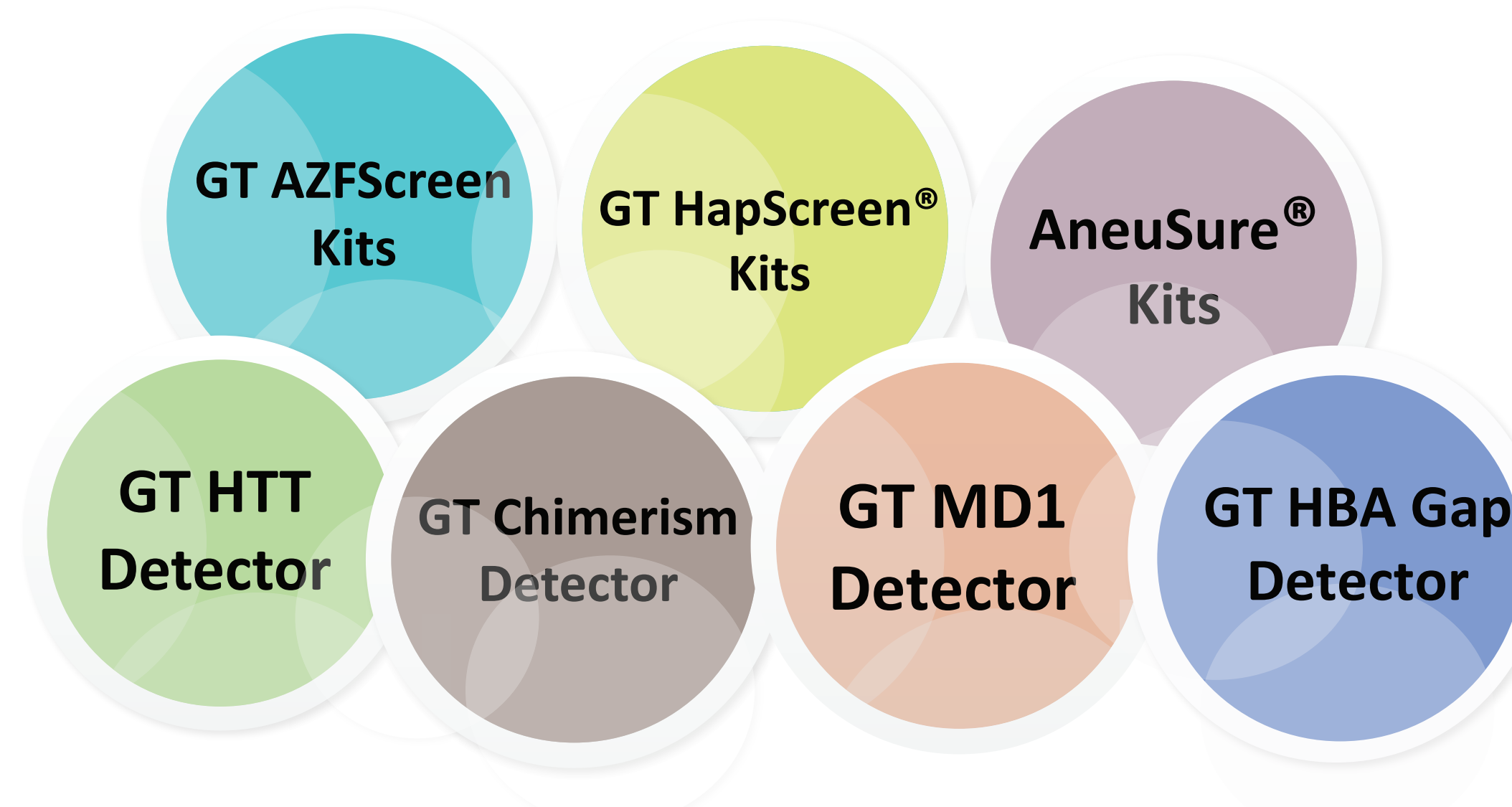
Summary of GT FA Detector's Key Features

Accurate quantification of GAA repeats using GT1200 Size Standard.

Compatible with Applied Biosystems® 3130/3130xl, 3500/3500xl and SeqStudio Genetic Analyzers.

Possibility of gel-based interpretation of results.

More from Genetek's Diagnostic Line



For further information please contact us via info@genetek.de

More details on Genetek products are available on our website: www.genetek-biopharma.com

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