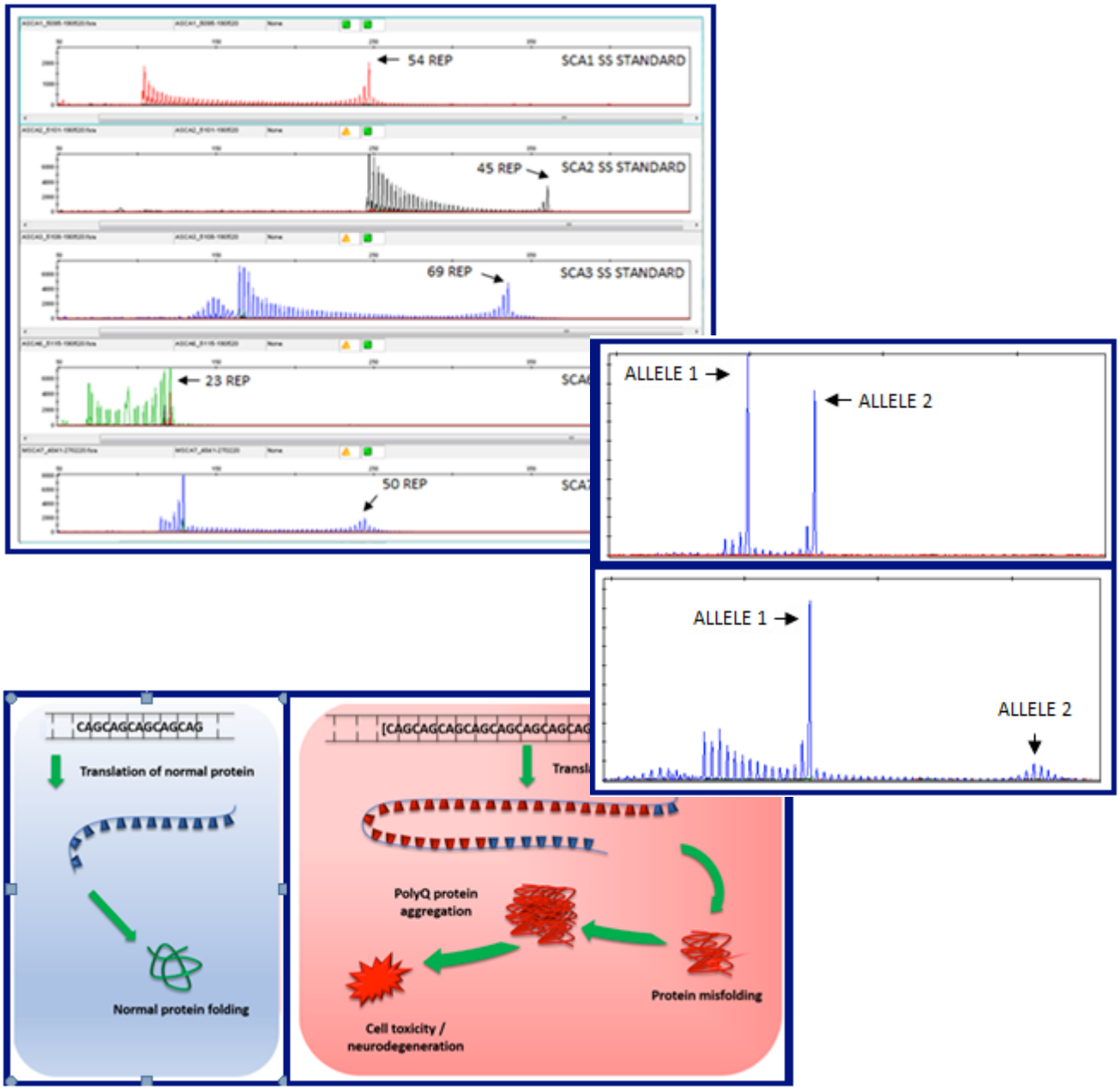
EXPERTEAM

l'esperienza in biologia molecolare

SYSTEM for the MOLECULAR ANALYSIS of SPINOCEREBELLAR ATAXIA type 1, 2, 3, 6, 7 by TP-PCR and CAPILLARY ELECTROPHORESIS

SCAs KIT-FL

cod. SC.01FL



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Autosomal dominant spinocerebellar ataxias (SCAs) are a group of clinically and genetically heterogeneous neurodegenerative diseases characterized by ataxic gait, slow progressing ataxia of posture and limbs, dysarthria and/or oculomotor disorders, caused by cerebellar degeneration in absence of other pathologies. The onset of the disease is generally between 20 and 40 years old.

To date, 47 SCA subtypes have been described; the prevalence is estimated at about 3/100,000 although some data refer to studies on restricted geographical areas. The founder effect helps to create distribution differences in various regions. The most common form in the world is SCA3 while in Italy the most common forms are SCA2 and SCA1.

The major subtypes of SCA are SCA1, SCA2, SCA3, SCA6 and SCA7 which together account for approximately 60% of the dominant autosomal spinocerebellar ataxias in the world. They are termed polyglutamine diseases in which CAG triplet expansions are located in a coding region of the gene resulting in abnormal elongation of polyglutamine (polyQ) in the corresponding protein. The predominant effect of these mutations is represented by a gain in function of aberrant proteins, favoring the ability to aggregate with other proteins

The SCAs KIT-FL is used for the amplification of the CAG polymorphic repetition contained in the genes *ATXN1, ATXN2, ATXN3, CACNA1A* and *ATXN7* and is used

in the molecular study of spinocerebellar ataxias types 1, 2, 3, 6 and 7.

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How does the kit work?

The SCAs kit-FL allows to identify all the pathogenic expansions involved in **SCA1, SCA2, SCA3, SCA6, SCA7**, to accurately determine the length of those below 80 CAG and to highlight any interruptions without resorting to second tests. level. The kit is based on the **TP-PCR** technique that makes use of a fluorescent primer that maps upstream of the repeated sequence and a chimeric reverse primer partially overlaid on the repeated region. The chimeric primer hybridizes at multiple sites within the repeated $CAG_{(n)}$ region, creating PCR products of various sizes. After the PCR reaction, the amplified products are separated by capillary electrophoresis. The interpretation of the results is based on the determination of the length of the previously amplified CAG triplet sequence and/or on the presence of "stutter" peaks in the electrophoretic trace.

The kit provides 5 amplification master mixes (each specific for a subtype of SCA) that allow the amplification of the repeated CAG regions contained in the *ATXN1, ATXN2, ATXN3, CACNA1A* and *ATXN7* genes through a single "touchdown" amplification program. The *SCA sequence specific standards* are supplied together with the kit which allow to accurately distinguish normal from pathological alleles.

Starting samples: peripheral blood

Thermal cyclers: T-professional (Biometra), Proflex PCR system/QS5 (Applied Biosystems).

DNA Sequencers: 3130/-XL, 3730/-XL, 3500/-XL and SeqStudio (Applied Biosystems).

Kit content

Label	Content
SCA 1,2,3,6,7 MASTER MIX	Mix for the amplification of SCA genes
SCA 1,2,3,6,7 SS STANDARD	Size standard containing alleles with known number of CAG repeats
SCA DNA polymerase	DNA polymerase for complex amplifications