

## SCA 1, SCA 2 & SCA 3/MJD mutations in ataxia syndromes in southern India

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**Background & objectives:** Spinocerebellar ataxias (SCAs) are often caused by expansions of CTG/CAG trinucleotide repeat in the genome. Expansions at the SCA1, 2 and 3 loci are the most frequent, but differences in their relative proportion in regions occur across the world. We carried out this study to assess the occurrence of SCA1, 2 and 3, at a tertiary neuro-psychiatric center in Bangalore, Karnataka.

**Methods:** Probands (N=318) who were diagnosed to have an ataxia syndrome (progressive degenerative ataxia of unknown cause) attending the clinical services of the National Institute of Mental Health and Neuro Sciences (NIMHANS), Bangalore, were evaluated over a period of three years. Standard protocols were used for both clinical and molecular diagnosis.

**Results:** Genotyping established that SCA1, 2 and 3 accounted for more than one third of the ataxia cases seen in the clinic. In the cases with established family history and autosomal dominant inheritance SCA1 was most prevalent followed by SCA2 and SCA3.

**Interpretation & conclusions:** Our findings suggested SCA1 rather than SCA2 to be the more common mutation in southern India. Large numbers of SCA3 probands were also identified. Differences in prevalence of these syndromes within India need to be explored further for founder effects, correlations with phenotype, and patterns of outcome. Family history was not apparent in almost a fifth of those tested positive, highlighting the value of testing even in the absence of family history. Molecular testing should be extended to cover the other forms of ataxia, of which a large number are now known. Combined efforts to confirm the presence of these less common forms, as well as family studies to detect novel mutations, are necessary in this context in India.

**Key words** Genetic markers - prevalence - spinocerebellar ataxia

Spinocerebellar ataxias (SCAs) are a group of neurological disorders, many caused by an expansion

of unstable (CAG) triplet repeats in the genome<sup>1</sup>. Evidence of anticipation, expansions of repeats in