Fragile X-associated tremor and ataxia syndrome (FXTAS) is an adult-onset neurodegenerative disorder that presents with symptoms associated with movement and cognition. These symptoms continue to worsen with age. Individuals with FXTAS have a premutation in which the CGG triplet repeats of the FMR1 gene is repeated 55 to 200 times compared to 5 to 40 times normally. However, only one third of male premutation carriers will clinically present with FXTAS. Researchers at Fred Hutch have identified a novel isoform of ASFMR1e that is transcribed anti-sense to the FMR1 locus. Transcript of this loci is upregulated in premutation carrier clinical samples and is highly expressed in human brain suggesting that alternative splice forms contribute to the variable clinical phenotypes associated with FXTAS.

### Applications
- Molecular diagnostic
- Patient stratification for clinical trials

### Advantages
- Molecular diagnostic biomarker to confirm premutation status of FMR1

### Market Overview
Approximately 1 in 450 males and 1 in 200 females have an FMR1 gene premutation, putting them at risk for FXTAS. However, not everyone with a premutation will show signs and symptoms of FXTAS. In those over age 50, with this premutation, 30-40% of males and 8-16% of females will develop FXTAS with prevalence increasing with age for men. In the general population, about 1 in 3,000 men over age 50 suffer from symptoms of FXTAS.

### Investigator Overview
Stephen Tapscott, MD, PhD, Human Biology Division