SPINAL AND BULBAR MUSCULAR ATROPHY (SBMA)
OMIM #313200

INTRODUCTION
SBMA (Kennedy disease) is a slowly progressive neuromuscular disorder resulting in proximal muscle weakness with involvement of the facial, neck and throat muscles. SBMA is inherited in an X-linked manner. Males may also demonstrate mild androgen insensitivity including gynaecomastia and/or small testes with oligo- or azoospermia. Females are generally unaffected (dependent on X-inactivation pattern and circulating male hormone levels). The prevalence of SBMA is estimated to be 1 in 40,000 - 50,000 live male births in populations of Western European descent. A trinucleotide (CAG)n repeat expansion in exon 1 of the Androgen Receptor gene (AR), located on Xq11-q12, is the only mutation associated with the disease. Other mutations within the AR gene are not associated with SBMA but are associated with more severe forms of androgen insensitivity.

TESTING AND REFERRALS
Diagnostic:
- For individuals suspected of having SBMA
- From Clinical Genetics, Clinical Neurology or Paediatric Neurology
Carrier:
- Asymptomatic mothers of affected sons
- Obligate carrier confirmation
- Daughters of affected males – can constitute presymptomatic tests and are treated as such
- From Clinical Genetics only
Presymptomatic:
- Referrals for asymptomatic individuals with clinically affected relatives
- Daughters of affected males
- From Clinical Genetics only
Prenatal:
- Prenatal testing must be discussed with the laboratory and arranged in advance
- From Clinical Genetics and Prenatal Diagnosis only

TECHNICAL INFORMATION AND STRATEGY
- PCR across the AR exon 1 (CAG)n repeat
- Sexing of foetal samples prior to (CAG)n repeat analysis

TARGET REPORTING TIMES
Diagnostic test: 10 days
Presymptomatic/carrier test: 10 days
Prenatal test (includes maternal contamination check): 3 days

N.B. Details are correct for the date of printing – last updated 01/09/2015