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  "name": "Human[Tax:9606]autosomal dominant ataxia [D0:0050954] SCA1. ",
  "title": "Spinocerebellar Ataxia Type 1 (SCA1)",
  "version": "1.0",
  "createdby": "amandab2140@gwmail.gwu.edu",
  "created": "Feb 02, 2017 11:11:00",
  "modified": "Feb 23, 2017 15:42:38",
  "digital_signature": "",
  "verification_status": "unreviewed",
  "publication_status": "draft",
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    "Updated Spinocerebellar Ataxia Type 1 (SCA1) BC0",
    "Determine CAG repeats [S0:000726] in a sequence for the human [TAX:9606] Spinocerebellar Ataxia Type 1 (SCA1), autosomal dominant ataxia [D0:0050954], implicated on ATXN1 gene [G0:0000037] 6p22.3 [OMIM:164400].Diagnosis for SCA1 results in the molecular genetic testing to detect abnormal CAG repeats on the ATXN1 gene.",
    "Criteria for Diagnosis: Alleles with less than 35 CAG repeats have not been associated with SCA1 phenotype [PMID:8634720].",
    "Normal: 36 – 44 CAG repeats in presence of CAT trinucleotide repeats that disrupts the CAG repeats. ",
    "Mutable–normal: 36 – 38 CAG repeats without CAT trinucleotide repeat disruptions.",
    "Full–penetrance: 39 and above CAG trinucleotide repeats."
  ],
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    "authors": [
      {"orchid": "0000-0003-1409-4549"},
      {"orchid": "0000-0002-9920-565X"}
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        "https://www.ncbi.nlm.nih.gov/nuccore/NM\_000332.3"
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    "false negative discovery < 0.10",  
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