Supplement 2  Search criteria systematic review

We reviewed all papers regarding dystonia, both genetic and non-genetic, in three age groups (infancy, childhood and adolescence), which is from birth to 20 years of age. The key terms we used were “dystonia” combined with (synonyms of) “children”, “childhood”, “adolescence” and “early onset”, as well as (synonyms of) terms indicating possible etiologies including “genetic”, “acquired”, “primary”, “secondary”, “heredodegenerative”, “hereditary”, and “inborn errors of metabolism”. All reviewed papers and abstracts were presented in English.

We considered using the Quality Assessment of Diagnostic Accuracy Studies (QUADAS-2) tool for selecting papers, developed by Whiting and colleagues (see Supplemental references). However, this tool proved not to be applicable as disorders causing dystonia of childhood are rare and the available evidence consisted only of small clinical trials, case series and expert opinion. For the same reason, not all items on the PRISMA Checklist (Supplement 6) are applicable.

Study selection: included literature and associated references involved at least one adequate description of: symptoms, signs, laboratory investigations (including metabolic evaluation), neuroimaging or genetic analysis, with supporting evidence for the etiological diagnosis. Included literature concerned at least two patients with the same condition, presenting with dystonia as an isolated, prominent or presenting symptom. Further references were retrieved manually from reference lists. Text books, Online Mendelian Inheritance in Man (OMIM) and GeneReviews were used for overviews of possible causes of dystonia. The list of genes (Supplement 1) is based on a detailed literature search up to October 20th 2014). The final reference list was generated on the basis of originality and relevance to the topic.
Key electronic search strategy for PubMed:

(dyston* AND (child* OR pediatric OR adolescen* OR (early onset) OR (early-onset))) AND
(genetic OR primary OR hereditary OR heredodegenerative OR acquired OR secondary OR
(inborn errors of metabolism))

Filters activated: Humans, English.