Table 4 Biochemical investigations to identify treatable inborn errors of metabolism with dystonia as an important feature

Laboratory test	In sample of	Disorder
Organic acids	Urine	Glutaric aciduria type I, propionic aciduria, methylmalonic aciduria, cobalamin deficiencies
Lactate	Plasma	Propionic aciduria, methylmalonic aciduria, biotin responsive basal ganglia disease
Pyruvate	Plasma	Pyruvate dehydrogenase complex deficiency
Acylcarnitines	Plasma	Propionic aciduria, methylmalonic aciduria, glutaric aciduria type 1
Amino acids	Plasma	Ornithine transcarbamylase deficiency, maple syrup urine disease, pterin defects
Homocysteine	Plasma	Homocystinuria
Copper, ceruloplasmin	Plasma, urine	Wilson's disease
Manganese	Plasma	Dystonia with brain manganese accumulation
Biotinidase	Plasma	Biotinidase deficiency
Creatine, guanidinoacetic acid	Plasma, urine	Cerebral creatine deficiency syndrome 3 (AGAT deficiency), guanidinoacetate methyltransferase deficiency
Vitamin E (α-tocopherol)	Plasma	Ataxia with vitamin E deficiency
Uric acid	Plasma	Lesch-Nyhan syndrome
Cholestanol	Plasma	Cerebrotendinous xanthomatosis
Glucose	CSF, plasma	GLUT-1 deficiency
Folate	CSF	Cerebral folate deficiency
HVA, 5-HIAA	CSF	Tyrosine hydroxylase deficiency
Pterines	CSF, urine	GTP-cyclohydrolase 1 deficiency, 6-pyruvoyl-tetrahydropterin synthase deficiency, aromatic l-amino acid decarboxylase deficiency
Sepiapterin	CSF	Sepiapterin reductase deficiency

Performing this set of laboratory investigations is only recommended if obtaining the results of these tests will be faster than NGS testing.
Lumbar puncture seems justified only in selected cases with a high clinical suspicion for these disorders.
AADC, aromatic l-amino acid decarboxylase; AVED, ataxia with vitamin E deficiency; CSF, cerebrospinal fluid; NGS, next-generation sequencing; PDC, pyruvate dehydrogenase complex; PTPS, 6-pyruvoyl-tetrahydropterin synthase.