

Signs and symptoms

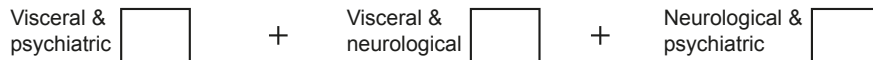
Indicators	Visceral	Score	Neurological	Score	Psychiatric	Score
Very strong 40 points per item			<ul style="list-style-type: none"> • Vertical supranuclear gaze palsy • Gelastic cataplexy 	<input type="checkbox"/> <input type="checkbox"/>		
Strong 20 points per item	<ul style="list-style-type: none"> • Prolonged unexplained neonatal jaundice or cholestasis • Isolated unexplained splenomegaly (historical and/or current) with/without hepatomegaly 	<input type="checkbox"/> <input type="checkbox"/>			<ul style="list-style-type: none"> • Pre-senile cognitive decline and/or dementia 	<input type="checkbox"/>
Moderate 10 points per item			<ul style="list-style-type: none"> • Ataxia, clumsiness or frequent falls • Dysarthria and/or dysphagia • Dystonia 	<input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/>	<ul style="list-style-type: none"> • Psychotic symptoms (hallucinations, delusions and/or thought disorder) 	<input type="checkbox"/>
Weak 5 points per item			<ul style="list-style-type: none"> • Acquired and progressive spasticity 	<input type="checkbox"/>	<ul style="list-style-type: none"> • Treatment-resistant psychiatric symptoms • Other psychiatric disorders 	<input type="checkbox"/> <input type="checkbox"/>
Ancillary 1 point per item	<ul style="list-style-type: none"> • Hydrops fetalis • Siblings with fatal ascites 	<input type="checkbox"/> <input type="checkbox"/>	<ul style="list-style-type: none"> • Hypotonia • Delayed developmental milestones • Seizure (partial or generalised) • Myoclonus 	<input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/>	<ul style="list-style-type: none"> • Disruptive or aggressive behaviour in adolescence and childhood 	<input type="checkbox"/>

Category scores



Category Combination

40 points: Visceral & psychiatric
 40 points: Visceral & neurological
 20 points: Neurological & psychiatrics



NP-C family relationship

40 points: Parent/sibling
 10 points: Cousin



Risk Prediction Score

= Sum of scores