Instruments for Assessment of Cognitive Function for **Children with Spinal Muscular Atrophy: A Targeted** Literature Review

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Introduction

- SMA is a congenital motor neuron disease leading to progressive muscle weakness, disability, and in the most severe cases without treatment, infantile death¹
- SMA is categorized into five clinical subgroups ranging from 0 (most severe) to 4 (least severe) based on the age of onset, severity of symptoms, highest motor milestone achieved, and genotype¹
- SMA type 1, accounting for >45% of SMA cases (life expectancy ≤2 years), is the most severe form of SMA in which patients have respiratory insufficiency, poor feeding, and are unable to sit independently¹
- SMA types 2 and 3 occur at a lower frequency (20% and 30% of cases, respectively) and have less severe clinical manifestations with patient life expectancies ranging from approximately 25 years of age up to a normal life span¹
- Although most SMA studies focus on motor function, evaluating cognitive function in patients with SMA via validated instruments could further elucidate SMA pathology and treatment effects^{2,3}
- There is an incomplete understanding of the cognitive impacts of SMA which may be attributed to:^{2,3}
- Difficulty of assessing cerebral function and cognitive ability in severely affected patients
- Difficulty assessing severely disabled children with alternatives to standardized tests (e.g., eye tracker) – Physical symptoms (e.g., muscle weakness and respiratory and bulbar dysfunction) limiting environmental interactions

Cognitive function instruments

• A total of 28 cognitive function instruments were identified from the included publications (Table 2)

• The most common instruments used included the WIS, Bayley, CBCL, eye tracking devices, PedsQL-FIM, and Raven's test (Figure 2)

Table 2. Cognitive function instruments utilized

Instrument (number of publications)	Version	SN	IA ty	ype	- Age group	
	VCISION	1	2	3		
Instruments used in two or more publications						
	WISC-R	1	1	1	6–16 y	
	HAWIK-R	1	1	1	12–15 y	
VIS (n=11) ^{14,10,19,24-26,32,36,38-40} VIS (n=11) ^{14,10,19,24-26,32,36,38-40} ayley (n=6) ^{5,7,8,15,25,31} ayley (n=6) ^{5,7,8,15,25,31} BCL (n=5) ^{20,28,00,7,39} BCL (n=5) ^{20,28,00,7,39} ye tracker devices (n=5) ^{3,12,14,16,17} edsQL-FIM (n=4) ^{23,33-35} acken's test (n=4) ^{13,32,39,40} aters's test (n=2) ^{14,16} ECLA TVP (language abilities) (n=1) ^{19,36} ECLA TVP (language test (n=2) ^{16,23} patial location memory test (n=2) ^{21,22} struments used in one publicationBAS-III ¹⁷⁷ attery for linguistic assessment ²⁶ ECLA ¹⁴⁴ test (memory) ³⁶ ECRQ ²⁹ rench language reading test and reading aloudexicogrammatical analysis (languageevicogrammatical analysis (languageevicogrammatical analysis (languageevicogrammatical analysis (languageevicogrammatical analysis (languageevicogrammatical analysis (languageevicogrammat	HAWIE-R	1	1	1	16–18 y	
	WIS-NR	\checkmark	✓		3 6-16 y 12-15 y ✓ 16-18 y ✓ 6-16 y and 11 ✓ 2 y, 6 m-7 y, 3r ✓ 1-42 m ✓ 1-42 m ✓ 1-42 m ✓ 4-18 y ✓ 11-18 y ✓ 4-18 y ✓ 5-11 y ✓ 6-16 y ✓ 11-18 y ✓ 11-18 y ✓ 11 -18 y ✓ 11 -18 y ✓ 11 -18 y ✓ 12 -12 y ✓ 12 -12 y ✓ 2.5 -12.5 y ✓ 3-12 m ✓ 3-30 m 2.5 -8.5 y 2.5 -8.5 y	
WIS (n=11) ^{14,16,19,24–26,32,36,38–40}	WISC-IV		1	1	6–16 y and 11 m	
	WAIS-R		1	1	≥16 y	
	WPPSI-III		1		2 y, 6 m–7 y, 3m	
	WAIS				≥16 y	
	WISC				5–15y	
D_{1} L_{2} (0.578152531	BSID-III	1	1	1	1–42 m	
Bayley (n=6) ^{3,7,0,13,23,31}	NR	1			1–42 m	
	Achenbach CBCL	1	1	1	4–18 y	
$OBOT (N=2)^{20,20,30,31,38}$	Achenbach Youth Self Report		✓		Age group 3 $($	
Eye tracker devices (n=5) ^{9,12,14,16,17}	NR	1	✓		Unknown	
PedsQL-FIM (n=4) ^{29,33–35}	FIM	1	✓	1	<18 y	
	CPM	1	✓	1	5–11 y	
Raven's test (n=4) ^{13,32,39,40}	SPM	1	✓	1	6–16 y	
	CPM and SPM	1	1	✓	Unknown	
	Griffith's Mental Development Scale	1	✓	✓	6-16 y and 11 n 2 y, 6 m-7 y, 3m $\geq 16 y$ $\geq 16 y$ $\geq 16 y$ $\leq 16 y$ $\leq 16 y$ $\leq -15 y$ $1-42 m$ $1-42 m$ $1-42 m$ $4-18 y$ $11-18 y$ $0 - 6 y and 1$ $4 - 18 y$ $4 - 16 y$ $0 - 6 y and 11 m$ $\leq 89 y$ $0 - 6 y and 11 m$ $\leq 89 y$ $0 - 16 y$ $0 - 16 y$ $0 - 16 y$ $0 - 16 y$ $0 - 10 y$ $6 - 16 y$ $0 - 10 y$ $6 - 10 y$ $0 - 10 y$ $6 - 10 y$ $0 - 11 y$ $6 - 12 y$ $3 - 12 y$ $0 - 11 y$ $6 - 15 y$ $3 - 12 y$ $10 - 11 y$ $6 - 15 y$ $3 - 12 m$ $18 m - 4 y$	
Griffith's Scale (n=3) ^{10,11,31}	Griffith's-III	1			0–6 y and 11 m	
BECLA TVP (language abilities) (n=1) ^{19,36}	NR				≤89 y	
CAS test (n=2) ^{14,16}	NR	1	✓		Unknown	
K-ABC (n=2) ^{32,39}	NR	1	✓	✓	2.5–12.5 y	
NEPSY (n=2) ^{14,16}	NEPSY-II	1	1		3–12 y	
Spatial language test (n=2) ^{16,23}	NR		✓		Unknown	
	NR	1	1		Unknown	
ABAS-III ³⁷	NR				≤89 y	
Battery for linguistic assessment ²⁶	NR		1	1	4–12 y	
BEM-144 test (memory) ³⁶	NR					
CERQ ²⁹	NR	1	1	1	-	
French language reading test and reading aloud test (reading index) ³⁶	NR				·	
IBQ-R ³⁰	NR	1	✓	1	3–12 m	
Lexicogrammatical analysis (language development) ¹⁸	NR		✓		18 m–4 y	
MB-CDI ⁵	NR	1			8–30 m	
MSCA ³⁶	NR				2.5–8.5 y	
PPVT ⁴⁰	PPVT-R	1	1		•	
Survey for language, communication, and	PPVT					
motor skills ⁴	NR					
TCGB ¹³	NR	1			≥3.5 y	
Test for speed and accuracy in reading, developed by the MT group AND Battery for the assessment of developmental reading and spelling disorders (reading abilities) ²⁶	NR		1		Unknown	
VABS ¹¹	VABS-II	1			≤90 y	
WeeFIM ²⁷	NR	1	1	1	6 m–7 y	
	INIX				011 — 7 y	

• The domains assessed most often in patients with SMA were language abilities and memory (**Table 3**)

Table 3. Domains evaluated in cognitive function instruments

(n=10) (n=3) <							
InstrumentVersionabilitiesMethod V (r=3)Version (r=3)performance (r=2)Bayley $BSID-III$ $[7,8,15,31]$ XXXNR $[5,25]$ XXXGriffith's Scale $Griffith's Hartal[10,11]XXXBattery for the linguistic assessmentNR[25]XXXBECLA TVPNR[19,36]XXXECLA TVPNR[19]XXXMB-CDINR[16]XXXMB-CDINR[16]XXXMSCANR[16]XXXStruey for language, communication, andmotor skilsNR[13]XXSpatial location memory testNR[13]XXXWeeFIMNRx[21,22]XXWisMR[21,22]XXXSpatial location memory testNRx[21,22]XXWisMiscaNRx[23,29]XXWisMiscaNRx[23,23]XXMiscaNRx[23,23]XXMiscaNRx[23,23]XXSpatial location memory testNRx[23,23]XWisMiscaNRx[23,23]XWisMiscaX[36,40][36,4$			Domains evalu	er of instrument	ents) [references]		
Bayley F3D-III [7,8,15,31] X X X Griffith's Scale Image: I	Instrument	Version	abilities			performance	
NR [5,25] X X X Griffith's Scale [10,11] X X X X Griffith's Scale [31] X X X X Battery for the linguistic assessment NR [26] X X X BECLATVP NR [19,36] X X X French language reading test and reading alcud test (reading index) NR [36] X X X MB-CDI NR [5] X X X X MSCA NR [36] X X X X MB-CDI NR [36] X X X X MSCA NR [36] X X X X MB-CDI NR [36] X X X X MSCA NR [36] X X X X GetBM-144 NR [21,21] X X X	Bayley	BSID-III	•	х	Х	Х	
Griffith's ScaleGriffith's Mental Development Scale[10,11]XXXBattery for the linguistic assessmentNR[26]XXXBECLATVPNR[19,36]XXXEECLATVPNR[19,36]XXXFrench language reading test and reading aloud test (reading index)NR[18]XXMB-CDINR[16]XXXMBCANR[16]XXXMSCANR[36]XXXSurvey for language, communication, and motor skillsNR[13]XXXBEM-144NRX[26]XXXKABCNR[13]XXXXSpatial location memory testNRX[21,22]XXVWSWISC-IVX[23,24][24,25][25]WISWISC-RXX[14,16],9,26,36,40]WISC-RXX[13,239]XXWISC-RXX[19,26,36,40][14,16],9,26,36,40]WISC-RXX[32,39]XX[25]WISC-RXX[32,39]X[25]WISC-RXX[32,39]X[25]WISC-RXX[32,39]X[25]WISC-RXX[22,39]X[25]WISC-RXX[23,39]X[2	Dayley	NR	•	х	Х	Х	
Confifting Mental Development Scale [31] X X X Battery for the linguistic assessment NR [26] X X X BECLA TVP NR [19,36] X X X EECLA TVP NR [19,36] X X X French language reading idex) NR [36] X X X Lexicogrammetical analysis NR [18] X X X MB-CDI NR [36] X X X MSCA NR [36] X X X MSCA NR [36] X X X MSCA NR [36] X X X Survey for language, communication, and motor skills NR [4] X X X EEM-144 NR [25,39] X X X X VeeFIM NR [21,22] X X X [40] X <td>Criffith's Scale</td> <td>Griffith's-III</td> <td>•</td> <td>Х</td> <td>Х</td> <td>Х</td>	Criffith's Scale	Griffith's-III	•	Х	Х	Х	
Battery for the linguistic assessment NR [26] X X X BECLATVP NR [19,36] X X X X French language reading test and reading aloud test (reading index) NR [36] X X X Lexicogrammatical analysis NR [18] X X X MB-CDI NR [5] X X X MB-CDI NR [5] X X X MB-CDI NR [36] X X X MSCA NR [36] X X X Survey for language, communication, and motor skills NR [4] X X X TCGB NR [13] X X X X BEM-144 NR [36] X X X K-ABC NR [21,22] X X X VeeFIM NR [27] [27] X X <td></td> <td></td> <td>-</td> <td>Х</td> <td>Х</td> <td>Х</td>			-	Х	Х	Х	
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MSCA NR [36] X X X Survey for language, communication, and motor skills NR [4] X X X TCGB NR [13] X X X X BEM-144 NR X [36] X X X Spatial location memory test NR X [32,39] X X WeeFIM NR X [27] [27] X WeeFIM NR X [23] Y X WeeFIM NR X [27] [27] X WeeFIM NR X [27] [27] X WiSC-IV PPVT-R X X [40] X WISC-IV X [23] [24] [24] [25] WISC-R X X [38] [38] [38] WISC-R X X [32] X X WISC-R X X<	Lexicogrammatical analysis	NR		Х	Х	Х	
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K-ABC NR X [32,39] X X Spatial location memory test NR X [21,22] X X WeeFIM NR X [27] [27] X PPVT PPVT-R X X [40] X Image: Spatial location memory test PPVT-R X X [40] X PPVT PPVT-R X X [40] X X Image: Spatial location memory test PPVT-R X X [40] X PPVT PPVT-R X X [23,24] [24,25] [25] WISC X X [38] [38] [38] [38] WISC-R X X [32,39] X [14,16,19,26,36,40] [14,16,19,26,36,40] [14,16,19,26,36,40] [14,16,19,26,36,40] [14,16,19,26,36,40] [14,16,19,26,36,40] [14,16,19,26,36,40] [14,16,19,26,36,40] [14,16,19,26,36,40] [14,16,19,26,36,40] [14,16,19,26,36,40] [14,16,19,26,36,40] [14,16,19,26,36,40	TCGB	NR	•	Х	Х	Х	
KABC NR X [32,39] X X Spatial location memory test NR X [21,22] X X WeeFIM NR X [27] [27] X X PPVT PPVT-R X X [40] X WISC-IV X [23,24] [24,25] [25] WISC X X [38] [38] WISC-R X X [19,26,36,40] [14,16,19,26, 36,40] HAWIE-R X X [32,39] X WISC-R X X [32,39] X Raven's test CPM X X [13,40]	BEM-144	NR	Х	√ [36]	Х	Х	
Spatial location memory testNRX $[21,22]$ XXWeeFIMNRX $[27]$ $[27]$ XPPVTPPVT-RXX $[40]$ XWISC-IVX $[23,24]$ $[24,25]$ $[25]$ WISCXX $[38]$ $[38]$ WISC-RXX $[19,26,36,40]$ $[14,16,19,26,36,40]$ HAWIE-RXX $[32,39]$ XHAWIK-RXX $[32,39]$ XRaven's testCPMXXXCPM and CPMXXX $[13,40]$	K-ABC	NR	Х		Х	Х	
WeeFIM NR X [27] [27] X PPVT PPVT-R X X $[40]$ X WISC-IV X [23,24] [24,25] [25] WISC X X [38] [38] WISC X X [38] [38] WISC-R X X [19,26,36,40] [14,16,19,26, 36,40] HAWIE-R X X [32,39] X HAWIK-R X X [32,39] X WPSI-III X X X [25] CPM X X X [13,40]	Spatial location memory test	NR	Х		Х	Х	
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$\operatorname{WISC-IV} \times [23,24] [24,25] [25]$ $\operatorname{WISC} \times \times \times [38] [38]$ $\operatorname{WISC-R} \times \times \times [19,26,36,40] [14,16,19,26, 36,40]$ $\operatorname{HAWIE-R} \times \times \times [32,39] \times (14,16,19,26, 36,40] \times (14,16,19,26, 36,40]$ $\operatorname{HAWIE-R} \times \times \times [13,2,39] \times (14,16,19,26, 36,40] \times (14,16,19,26, 36,40]$ $\operatorname{HAWIE-R} \times \times \times [13,2,39] \times (14,16,19,26, 36,40] \times (14,16,19,26,36,40] \times (14,16,19,10,10,10) \times (14,16,1$	PPVT	PPVT-R	Х	Х	-	Х	
$\begin{array}{c ccccccccccccccccccccccccccccccccccc$		WISC-IV	Х				
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$\begin{array}{c c c c c c c c c c c c c c c c c c c $		WISC-R	Х	Х	√ [19,26,36,40]		
HAWIK-R X X [32,39] X WPPSI-III X X X [25] CPM X X X [25] Raven's test CPM X X X		HAWIE-R	Х	Х		Х	
WPPSI-III X X X [25] CPM X X X [13,40]		HAWIK-R	Х	х	•	Х	
Raven's test		WPPSI-III	Х	х	Х		
	Deven's tast	CPM	Х	х	Х		
	Raven's test	CPM and SPM	Х	Х	Х	/ [32,39]	

- Limitations in communication needed to study cognitive and verbal skills
- Lack of validated and/or standardized measures to assess cognitive function in patients with SMA
- In studies evaluating cognitive function, there has been substantial debate regarding the presence/severity of cognitive impairment for patients with SMA, with some studies reporting normal cognitive development and others demonstrating impairment in cognitive function^{2,3}
- The variability in these outcomes may be attributed, in part, to variations in the cognitive function instruments used²

Objective

• We sought to identify cognitive function instruments used in studies including children and adolescents with SMA and to critically appraise the common and unique of aspects of cognitive functioning operationalized in measures used across SMA studies

Methods

 A TLR of English-language original research publications was conducted using EMBASE and PubMed (conducted on August 14 and August 22, 2023, respectively) with the PICOS criteria and database search strings listed in **Table 1** Additional bibliographic/hand searches were also conducted on September 1, 2023 Table 1. Criteria for study inclusion and exclusion and database search strings

Criteria	
Population	Infants and adolescents with SMA types 1, 2, or 3
Interventions	Any
Comparator	Any
Outcome	Cognitive function assessments, including assessments of language, memory, verbal IQ, non-verbal/ performance IQ, behavior, and clinical assessments (e.g., eye tracking) completed by patients with SMA or their parent/proxy reporter
Study design	Conducted in humans Clinical trials, observational studies, registry-based studies, case series, surveys, or interviews
Language	English
Geography	Any
Publication year	Any
Publication type	Journal articles with full-text publications or congress abstracts
Database search s	trings ^a
	1 'spinal muscular atrophy':ti,ab OR 'muscular atrophy':ti,ab OR (sma:ti,ab AND atrophy:ti,ab)
	 'cognition'/exp OR cognition OR cognitive OR 'cognitive impairment' OR verbal OR executive OR behavior* OR behaviour* OR language OR visuospatial OR 'attention memory' OR attention OR memory OR spatial OR reading OR arithmetic OR learning OR intelligence
EMBASE	3 #1 AND #2
	4 #3 AND [humans]/lim
	5 #4 AND [English]/lim
PubMed	 cognition OR cognitive OR 'cognitive impairment' OR verbal OR executive OR behavior* OR behaviour* OR language OR visuospatial OR 'attention memory' OR attention OR memory OR spatial OR reading OR arithmetic OR learning OR intelligence
	2 ("spinal muscular atrophy"[Title/Abstract]) OR ("muscular atrophy"[Title/Abstract]) OR (sma[Title/ Abstract] AND atrophy[Title/Abstract])
	 3 (cognition OR cognitive OR 'cognitive impairment' OR verbal OR executive OR behavior* OR behaviour* OR language OR visuospatial OR 'attention memory' OR attention OR memory OR spatial OR reading OR arithmetic OR learning OR intelligence) AND (("spinal muscular atrophy"[Title/Abstract]) OR ("muscular atrophy"[Title/Abstract]) OR (sma[Title/Abstract] AND atrophy[Title/Abstract]))
	4 #3 AND applied Humans Filter
	5 #4 AND applied English Language Filter

Cognitive function instruments listed in order of decreasing frequency of domain evaluation

BECLA TVP, Batterie d'Evaluation du langage Test de Vocabulaire Actif et Passif; BEM-144, Batterie d'Efficience Mnésique-144; BSID-III, Bayley Scales of Infant and Toddler Development, 3rd edition: CPM, colored progressive matrices: HAWIE-R, Hamburg-Wechsler Intelligence Test for Adults-Revised: HAWIK-R, Hamburg-Wechsle Intelligence Tests for Children–Revised; K-ABC, Kaufman Assessment Battery for Children; MB-CDI, MacArthur-Bates Communicative Development Inventories; MSCA MacCarthy Intelligence Scale for Children; NR, not reported; PPVT-(R), Peabody Picture Vocabulary Test, Revised; SPM, standard progressive matrices; TCGB, Test of Grammatical Comprehension for Children; WeeFIM, Functional Independence Measure for Children; WIS, Wechsler Intelligence Scale; WISC-(IV), Wechsler Intelligence Scale for Children. 4th edition: WISC-(R). Wechsler Intelligence Scale for Children–Revised: WPPSI-III. Wechsler Preschool and Primary Scale of Intelligence. 3rd edition

SMA, spinal muscular atrophy

^aResults from Embase and PubMed searches were combined and augmented by an additional bibliographic/hand search to identify relevant literature.

- All records retrieved from the TLR were screened based on the title and abstracts by one reviewer at the first-level screening. Abstracts that did not match the inclusion criteria and duplicate abstracts were excluded.
- During a second-level screening, eligibility criteria were applied to the full-text citations by one reviewer
- Data presented in the included publications were extracted by one reviewer using a structured data abstraction form that included the name and version of cognition instrument used, applicable age group, SMA type, content/domains, and findings ata

 An independent review conducted a random quality check of the extracte 	d data
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Cognitive function instruments listed in order of decreasing frequency of use

ABAS-III, Adaptive Behavioral Assessment System, 3rd edition; Bayley, Bayley Scales of Infant and Toddler Development; BECLA-TVP, Batterie d'Evaluation du langage Test de Vocabulaire Actif et Passif; BEM-144, Batterie d'Efficience Mnésique-144; BSID-III, Bayley Scales of Infant and Toddler Development, 3rd edition; CAS test, Chimeric Animal Stroop test; CBCL, Child Behavioral Checklist; CERQ, cognitive emotion regulation questionnaire; CPM, colored progressive matrices; HAWIE-R, Hamburg-Wechsler Intelligence Test for Adults-Revised; HAWIK-R, Hamburg-Wechsler Intelligence Tests for Children-Revised; IBQ-R, Infant Behavior Questionnaire Revised; K-ABC, Kaufman Assessment Battery for Children; m, months; MB-CDI, MacArthur-Bates Communicative Development Inventories; MSCA, MacCarthy Intelligence Scale for Children; NEPSY-(II), A Developmental Neuropsychological Assessment, 2nd edition; NR, not reported; PedsQL-FIM, Pediatric Quality of Life Inventory-Family Impact Model; PPVT-(R), Peabody Picture Vocabulary Test-(Revised); SMA, spinal muscular atrophy; SPM, standard progressive matrices; TCGB, Test of Grammatical Comprehension for Children; VABS-(II), Vineland Adaptive Behaviour System-(2nd edition); WAIS-(R), Wechsler Adult Intelligence Scale-(Revised) WeeFIM, Functional Independence Measure for Children; WIS, Wechsler Intelligence Scale; WISC-(IV), Wechsler Intelligence Scale for Children, 4th edition; WISC-(R), Wechsler Intelligence Scale for Children–(Revised); WPPSI-III, Wechsler Preschool and Primary Scale of Intelligence, 3rd edition; y, years.

Figure 2. Cognitive function instruments utilized in two or more publications

Limitations

- Studies included for data abstraction were limited to English-language publications only
- Cognitive instruments were not consistently applied across studies and included domains unrelated to cognition (such as health-related QoL)
- Studies often grouped together SMA types, though cognitive impairment may be more prevalent for patients with more severe types

Conclusions

 This TLR identified 37 publications in which cognitive function was evaluated in patients with SMA, highlighting the limitations of current instruments used to comprehensively capture the cognitive effects of SMA

 Studies utilized a wide array of cognitive function instruments that mainly assessed language abilities and memory in patients with SMA

• Overall, the literature suggests that there is neither a clear consensus nor any validated cognitive function instruments for the assessment of cognitive function in patients with SMA

Implications for SMA Research

- Assessment of cognition in the first years of life, when the neural foundations of cognition are forming, is inherently challenging
- While available instruments include domains related to cognitive function, no particular validated instrument is optimal for assessing cognition in patients with SMA
- The development and validation of a new SMA cognitive function instrument, tailored to the patient's age and SMA type, may help address this unmet need

References

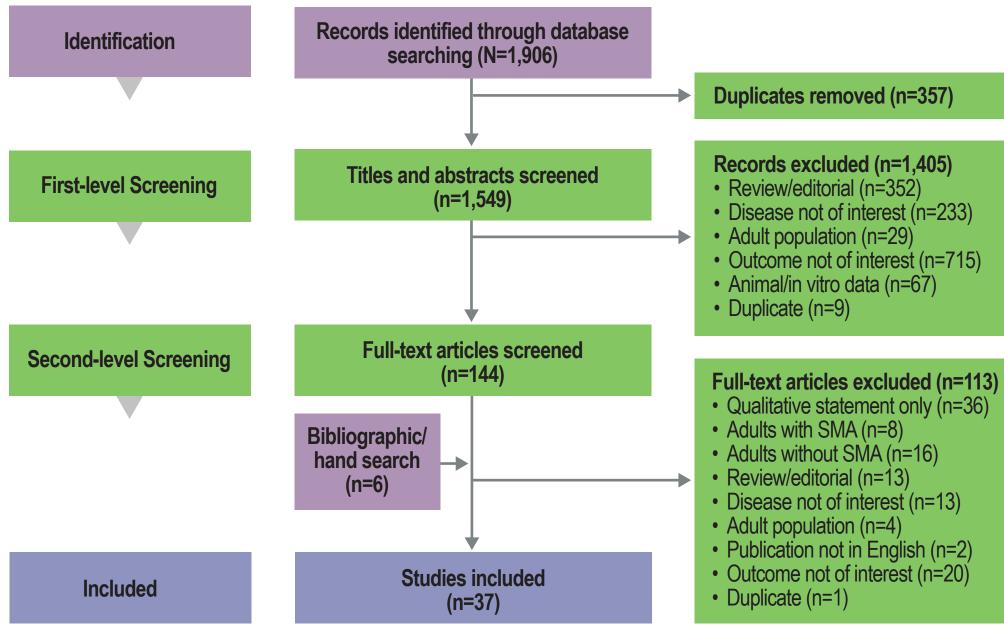
- 1. Keinath MC, et al. Appl Clin Genet. 2021;14:11–25.
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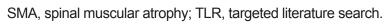
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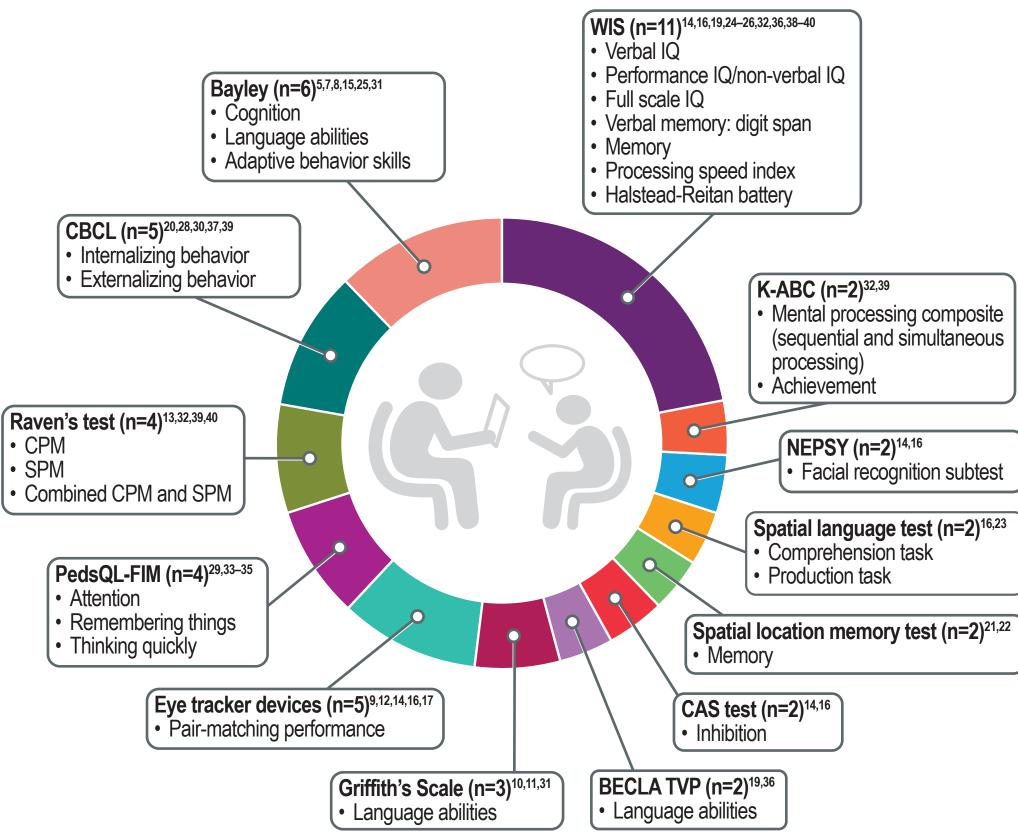
Studies

• The database search retrieved 1,906 records (Figure 1) • After screenings and a bibliographic/hand search, 37 publications were identified for data abstraction^{4–40}

Figure 1. TLR flow diagram







Bayley, Bayley Scales of Infant and Toddler Development; BECLA TVP, Batterie d'Evaluation du langage Test de Vocabulaire Actif et Passif; CAS test, Chimeric Animal Stroop test; CBCL, Child Behavioral Checklist; CPM, colored progressive matrices; PedsQL-FIM, Pediatric Quality of Life Inventory-Family Impact Model; K-ABC, Kaufman Assessment Battery for Children: NEPSY A Developmental Neuropsychological Assessment: SPM, standard progressive matrices; WIS, Wechsler Intelligence Scale

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Abbreviations

Bayley, Bayley Scales of Infant and Toddler Development; CBCL, Child Behavioral Checklist; DMD, Duchenne Muscular Dystrophy; PedsQL-FIM, Pediatric Quality of Life Inventory-Family Impact Model; PICOS, Population, Intervention, Comparison, Outcome, Study design; SMA, spinal muscular atrophy; TLR, targeted literature review; QoL quality of life; WIS, Wechsler Intelligence Scale.

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