Mental health in children with a neurogenetic syndrome associated with intellectual disability

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The Disability Paradox *Albrecht 1982*

Intellectual Disability

- Impairments
  - Physical comorbidities
  - Mental ill health and challenging behaviours
  - Poor sleep
- Functioning
  - Independence in daily living
  - Communication and mobility
  - Social skills

Outcomes

QUALITY OF LIFE
THRIVING
POSITIVE HEALTH
ENGAGEMENT
PRODUCTIVITY

Supports

- Physical and mental health
- Participation
- Maternal and family function
- Workplace functioning

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Children with disability can achieve a good quality of life

Genetic antecedents

Environmental determinants

Child health, wellbeing & participation

Family health and wellbeing

Impacts of intervention/supports

Eg,
Gastrostomy, scoliosis
Early intervention
Standing wheelchairs
Physical activity
Clinical trials – Rett, CDKL5 & Prader-Willi syndromes

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DEVELOPMENTAL MEDICINE & CHILD NEUROLOGY SYSTEMATIC REVIEW

The prevalence of mental health disorders and symptoms in children and adolescents with cerebral palsy: a systematic review and meta-analysis

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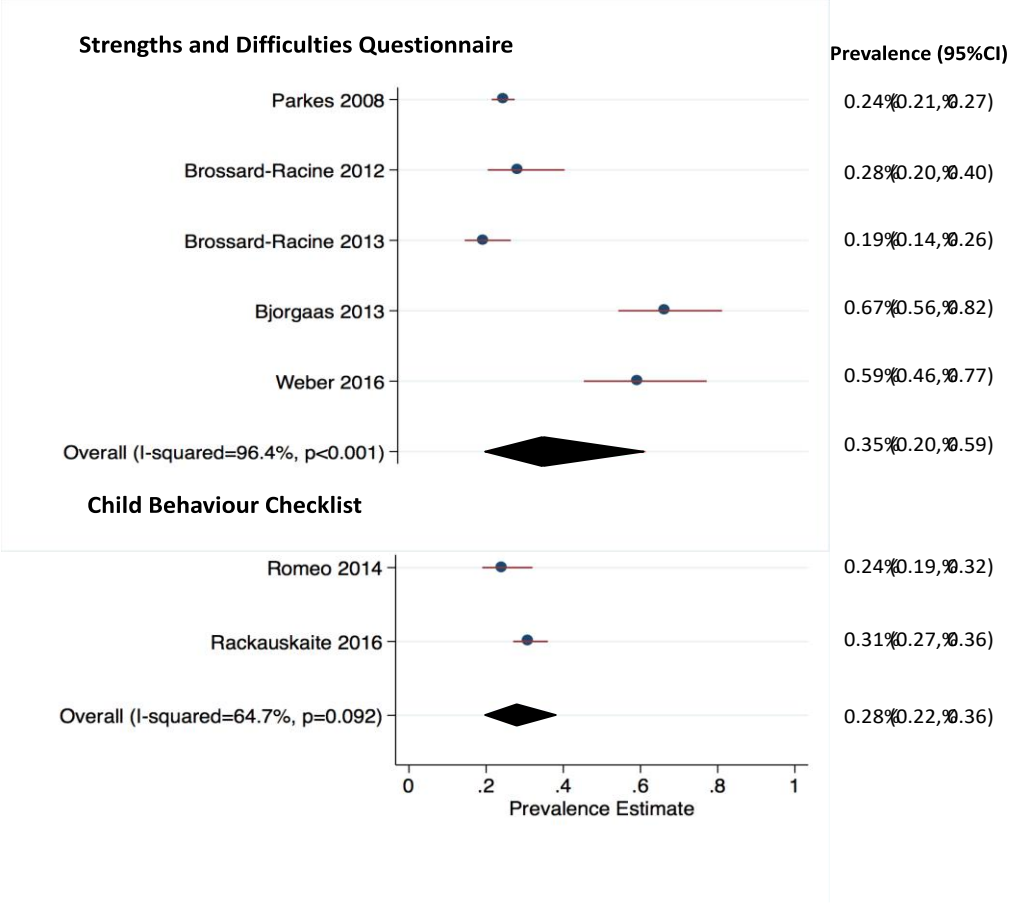
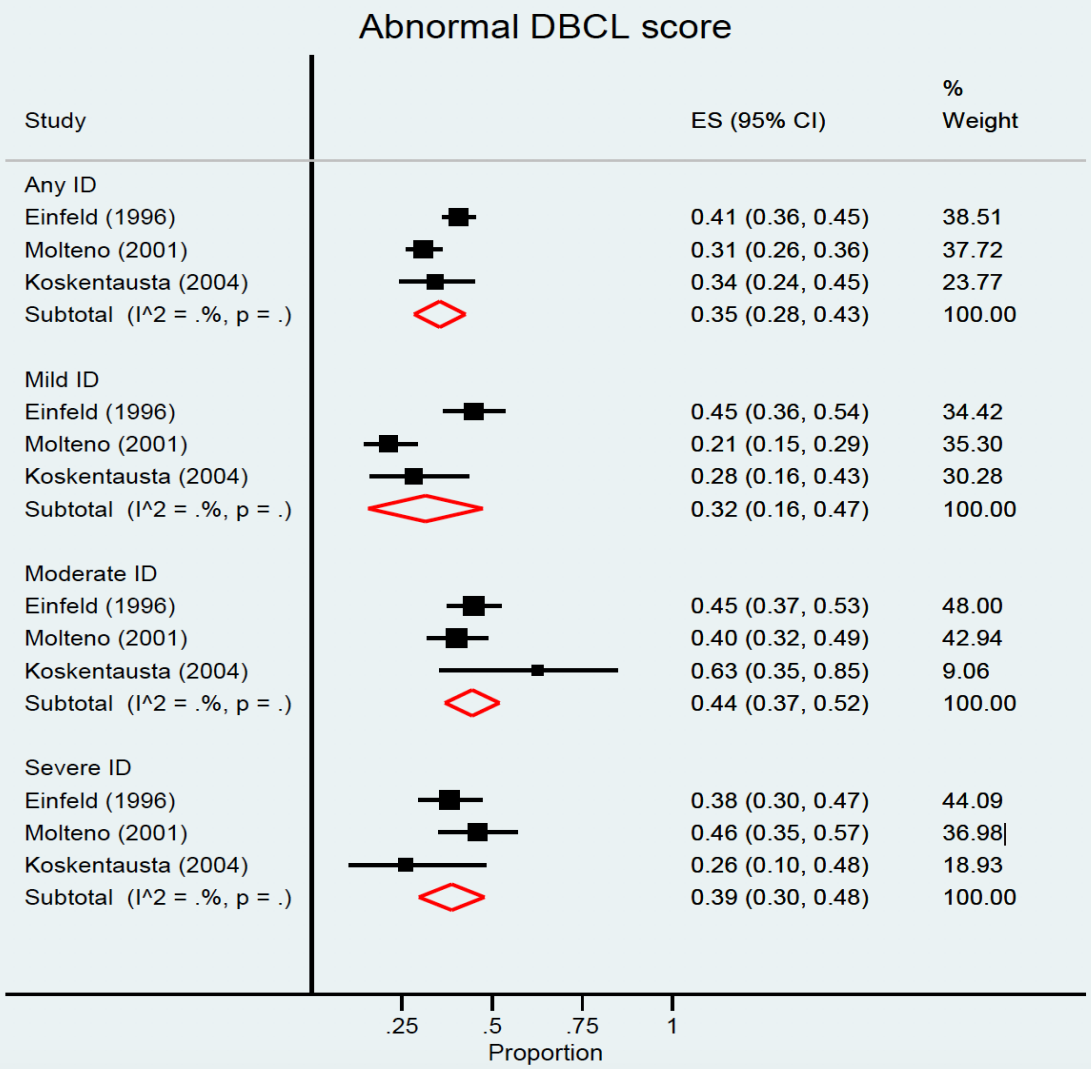


Title: Prevalence of mental health problems in children and adolescents with intellectual disability: a systematic review



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Title: Estimating prevalence of mental ill health in children with neurogenetic disorders: a systematic review

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The problem ….

- Mental health problems can be part of a syndromic gestalt
- Behavioural phenotypes vary distinctively across syndromes
  - Communication, daily activity, social activities, motor skills
- Mental health may also vary distinctively
- Propose that monogenetic disorders are valuable models

The method …

- Systematic review – 6-18y children
- 2,301 papers distilled to 40 papers investigating 4,241 children and adolescents with 1 of 10 syndromes
  - 7 studies with diagnostic interviews
  - 33 parent or teacher reported symptoms
  - 5 syndromes predominant - Down syndrome, 22q11.2 deletion syndrome, Fragile X syndrome, Williams syndrome and Prader-Willi syndrome
- CBCL and DBCL most common measures of symptoms

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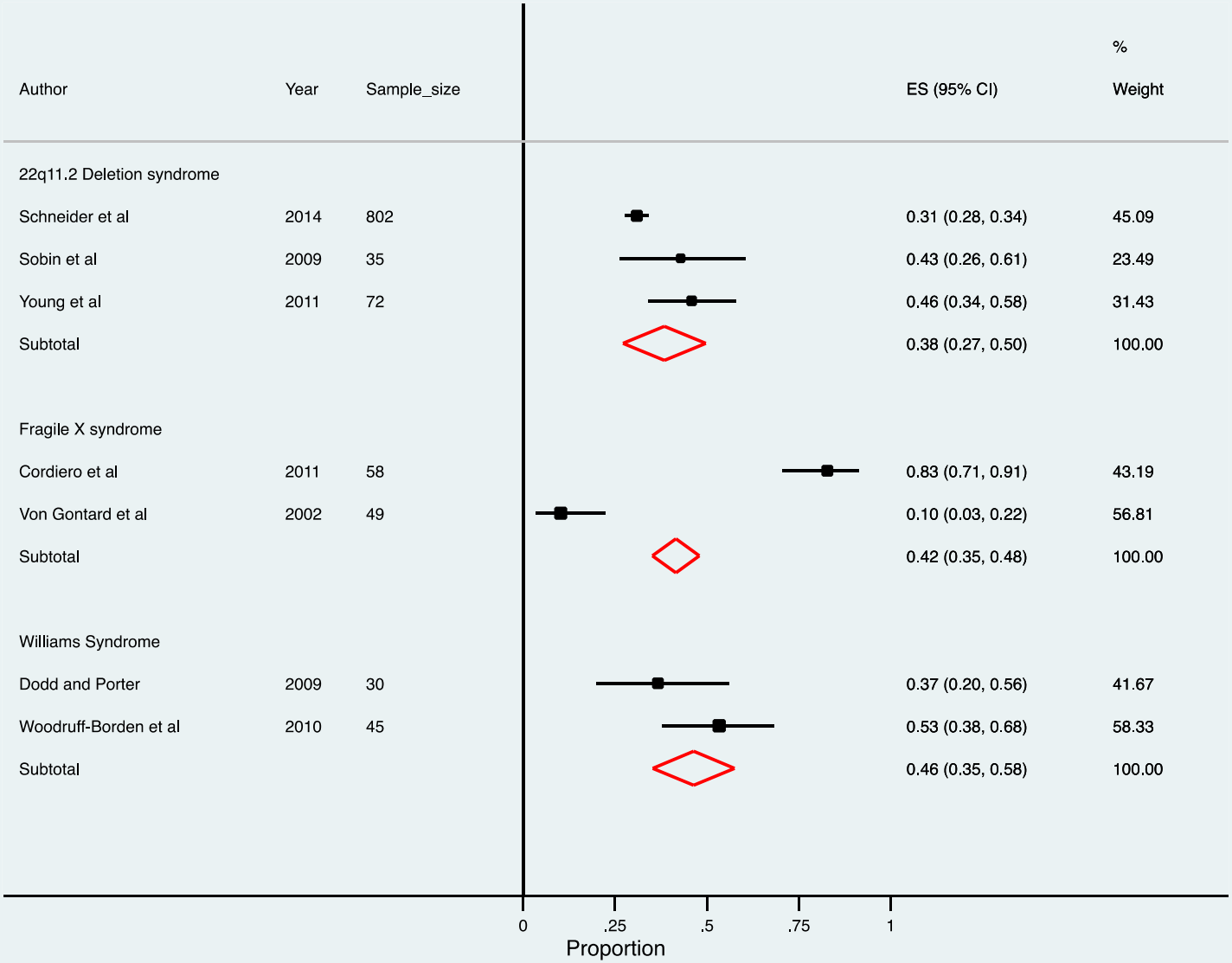
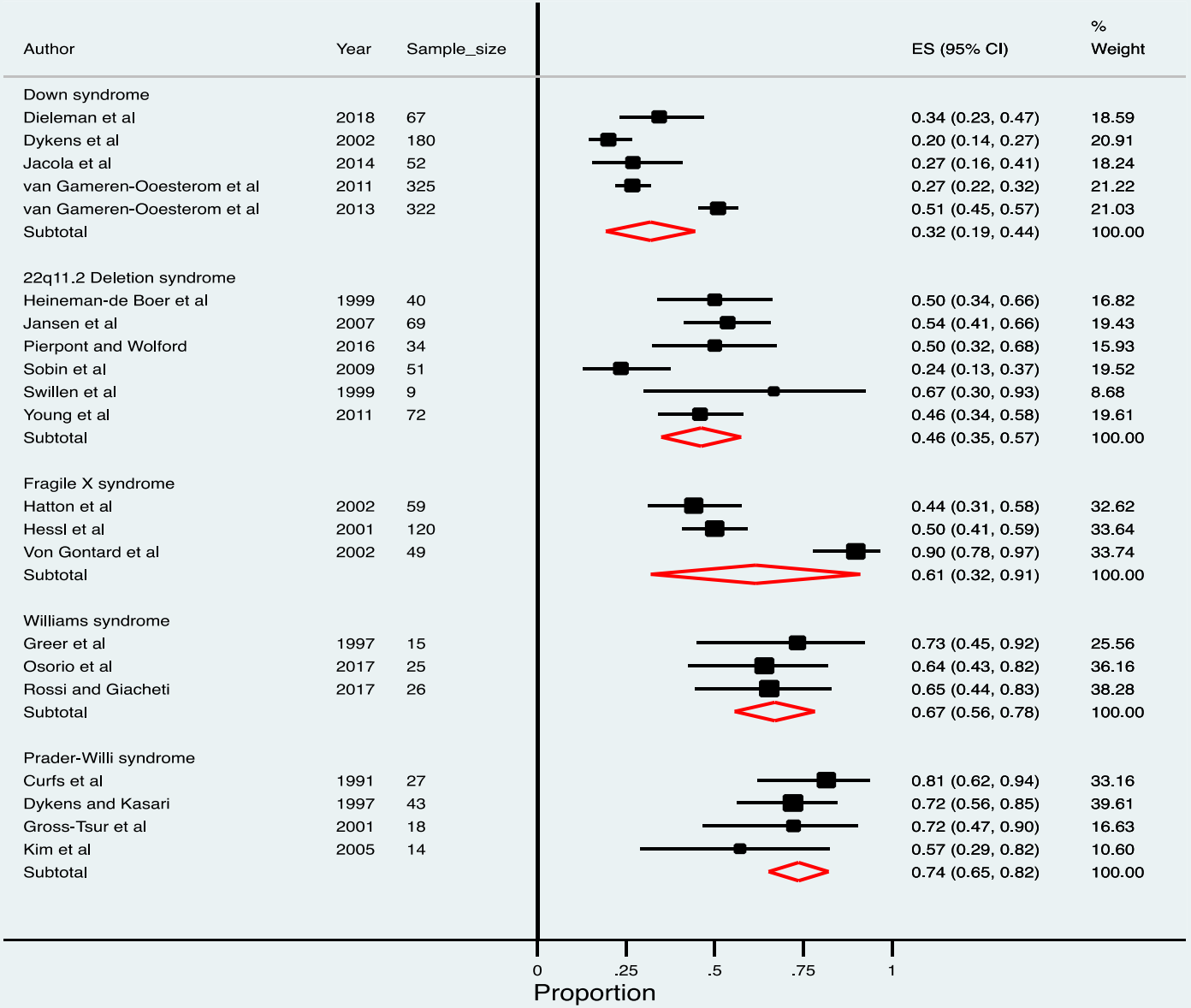
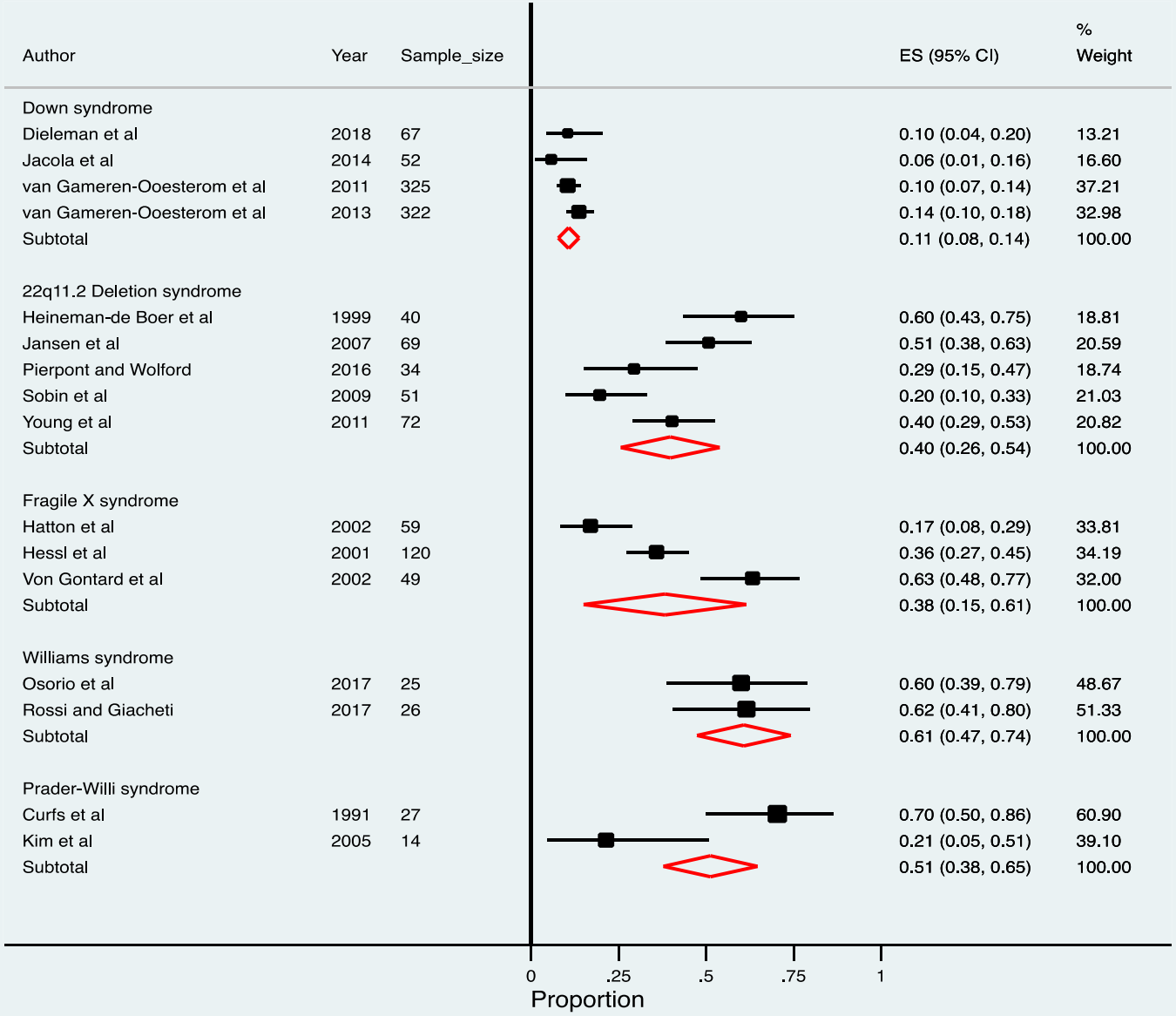
Diagnosis of Any Anxiety Disorder for select neurogenetic syndromes



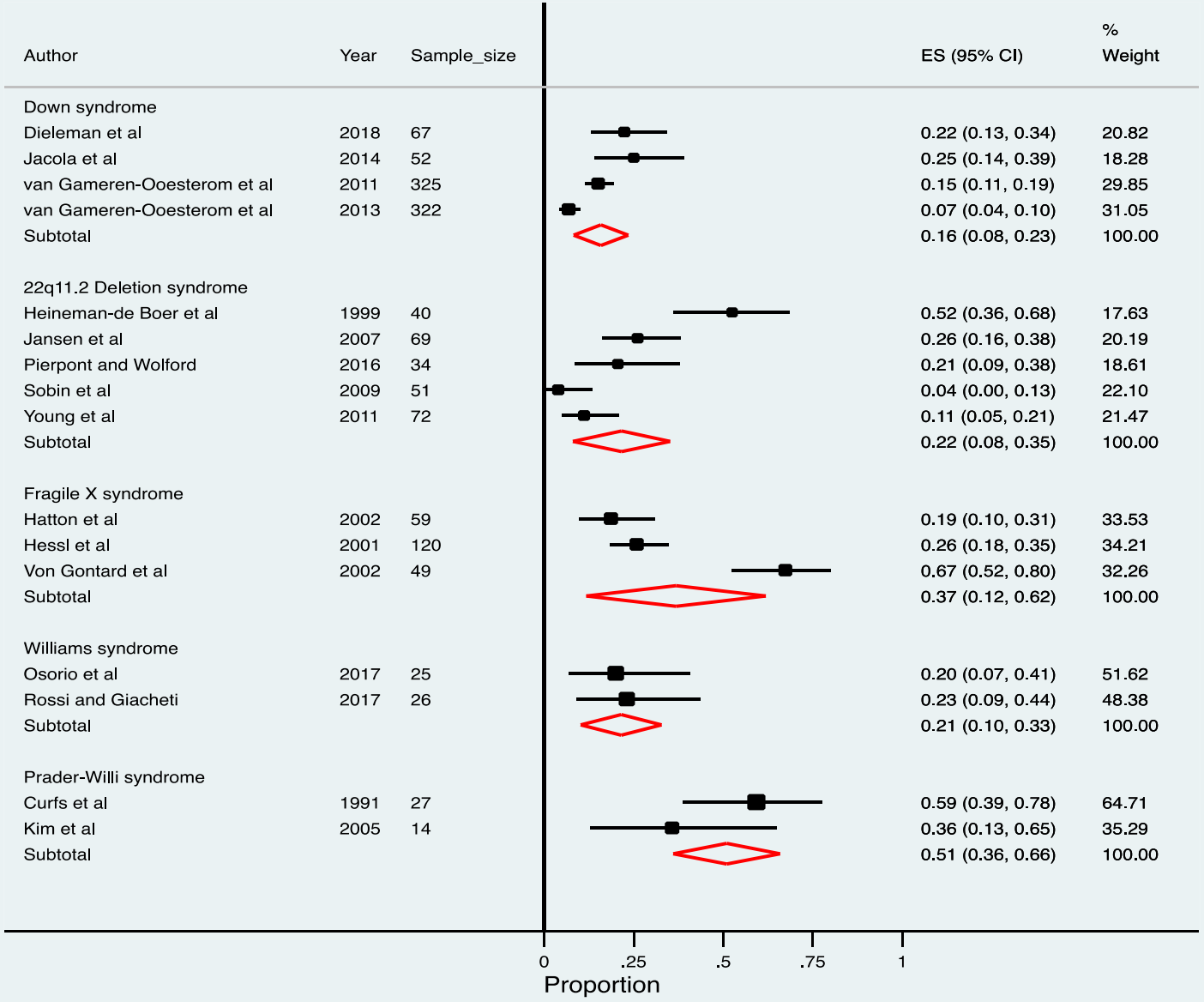
CBCL (Total Problem Behavior) scores for five neurogenetic syndromes



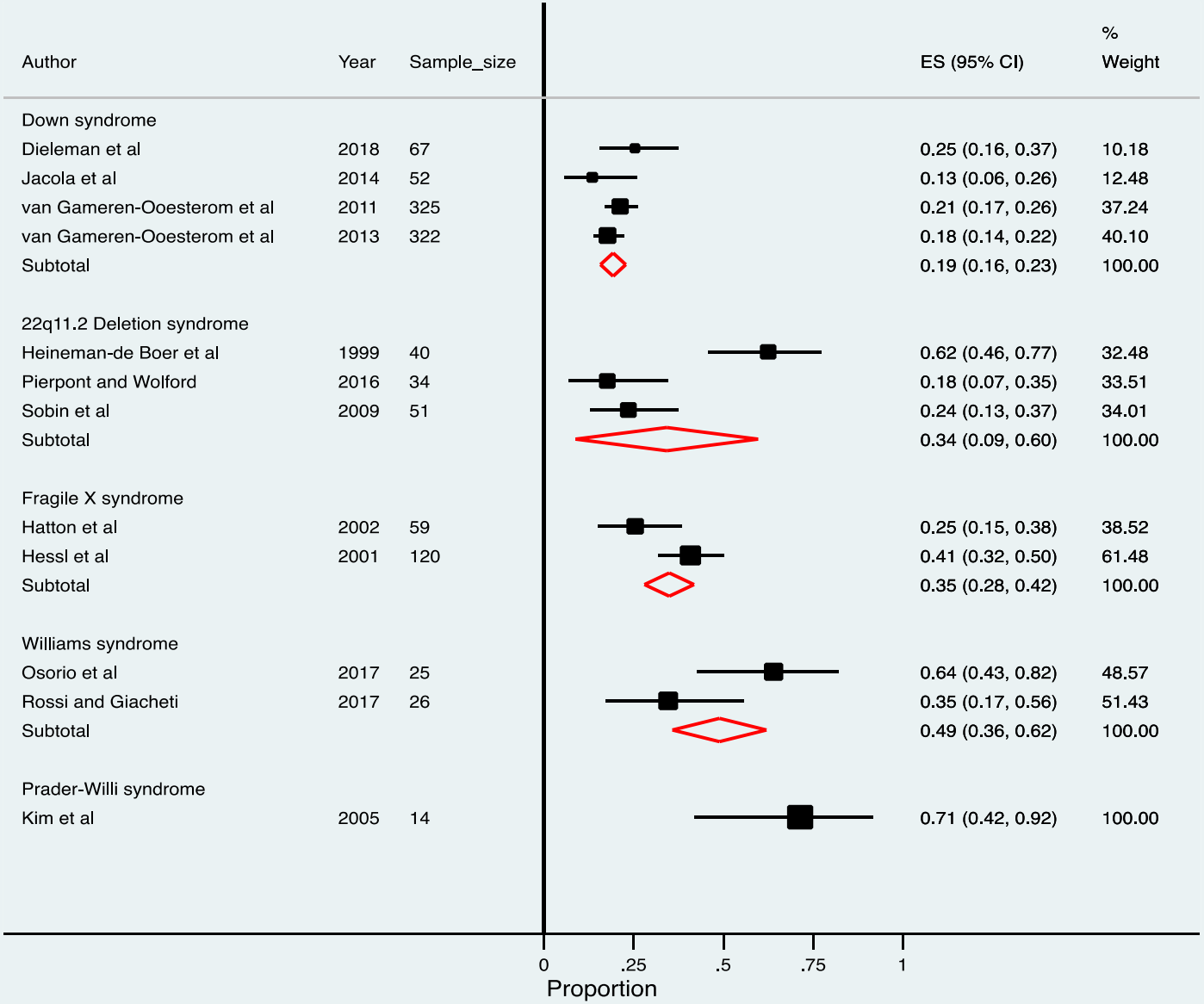
CBCL (Internalizing) scores for five neurogenetic syndromes



CBCL (Externalizing) scores for five neurogenetic syndromes



CBCL (Social Problems) scores for five neurogenetic syndromes



Comparison of CBCL scores with the general population

	Total score		Internalising domain score		Externalising domain score	
	Pooled prevalence % (95% CI)	P value[a]	Pooled prevalence (95% CI)	P value[a]	Pooled prevalence (95% CI)	P value[a]
General population	14.1 (13.0, 15.2)	Reference	12.8 (11.8, 13.8)	Reference	12.9 (11.9, 13.9)	Reference
Down syndrome	32.0 (19.0, 44.0)	0.004	11.0 (8.0, 14.0)	0.263	16.0 (8.0, 23.0)	0.392
22q11.2 Deletion	46.0 (35.0, 57.0)	<0.001	40.0 (26.0, 54.0)	<0.001	22.0 (8.0, 35.0)	<0.001
Fragile X syndrome	61.0 (32.0, 91.0)	<0.001	38.0 (15.0, 61.0)	0.032	37.0 (12.0, 62.0)	0.031
Williams syndrome	67.0 (56.0, 78.0)	<0.001	61.0 (47.0, 74.0)	<0.001	21.0 (19.0, 33.0)	<0.001
Prader-Willi syndrome	74.0 (65.0, 82.0)	<0.001	51.0 (38.0, 65.0)	<0.001	51.0 (36.0, 66.0)	<0.001

Discussion

- Prevalence higher than general population
  - CBCL total – 14% vs 32% (DS) to 74% (PWS)
- Differential vulnerability –
  - Partial specificity in phenotype
  - Syndrome-specific genetic contributions
- Higher levels of social ability or competence appeared to offer protection
  - Suggests strategies for more socially supportive environments
  - Eg, reduce loneliness, difficulties making friends and bullying

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Limitations

- Dataset included few papers, small sample sizes
  - Consortia for 22q11.2 syndrome
- Poor coverage of many neurogenetic disorders
- Unable to determine relationships between mental ill-health and ID severity level
- More studies needed on
  - Psychiatric diagnosis
  - Linkages between child and parental mental health
  - Socially supportive environments
  - Biological pathways

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What can we do to support mental health in children with ID?

Observational studies:

- Trajectories for child and parental mental health
- Mental health service use

Intervention studies:

- Preventive – child and parent
- Home, NDIS and education settings
- Clinical trials - ? Adaptive trials
- Best practice models
- Implementation science

Mental health literacy for ID and clarity of assessment and treatment pathways:

- Parents
- Front line practitioners
- Social

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