Study	Reference	Number of patients	Number of MR CNVs	Group	Targets	Number of targets	Mean spacing of targets	Follow-up studies
1	Vissers *et al*. (*1*)	20	2	MR and additional dysmorphisms (Checklist score >3)*	BAC	3,569	1 Mb	FISH
2	Shaw-Smith *et al*. (*2*)	50	7	Learning disability and dysmorphisms	BAC	3,500	1 Mb	FISH, genotyping
3	de Vries *et al*. (*3*)	100	11	MR with/or without congenital malformations and/or dysmorphism	BAC	32,447	100 kb	FISH, MLPA
4	Schoumans *et al*. (*4*)	41	4	MR (mild to severe) and dysmorphic features, malformations, and/or a family history (Checklist score >3)*	BAC	2,600	1 Mb	FISH
5	Tyson *et al*. (*5*)	22	2	MR (mild to severe) and nonsyndromic pattern of dysmorphic features	BAC	1,003 or 2,600	1 Mb, 3 Mb	FISH, microsatellite DNA analysis, real-time PCR
6	Menten *et al*. (*6*)	140	12	MR with one or more major congenital malformation or dysmorphism, or both	BAC	3,431	1 Mb	FISH, RT qPCR
7	Miyake *et al*. (*7*)	30	2	MR associated with some dysmorpic features	BAC	2,173	1.5 Mb	FISH
8	Friedman *et al*. (*8*)	100	10	MR (moderate-severe) and > additional clinical features: one major malformation, microcephaly, abnormal growth, or multiple minor anomalies	oligo	100,000	30 kb	FISH
9	Krepischi-Santos *et al*. (*9*)	95	13	Syndromic phenotype; almost all patients presented MR	BAC	3500	1 Mb	FISH, MLPA
10	Rosenberg *et al*. (*10*)	81	7	MR (mild to severe) and cranial/facial dysmorphisms and at least one additional dysmorphic feature	BAC	3500	1 Mb	FISH, MAPH
11	Engels *et al*. (*11*)	60	3	MR mostly combined with congenital anomalies	BAC	6000 or 8000	500 kb	FISH
12	Hoyer *et al*. (*12*)	104	10	MR, with or without multiple congenital anomalies	oligo	100,000	30 kb	FISH, qPCR for selected deletions <100kb and dup
13	Fan *et al*. (*13*)	100	6	MR (Checklist >3)*	oligo	44,290	30-35 kb	FISH, BaCGH or OaCGH244K;
14	Aradhya *et al*. (*14*)	20	10	MR and dysmorphic features or congenital anomalies or growth retardation	oligo, BAC	44,290, 2,600	30-35 kb, 1 Mb	FISH
15	Svensson *et al*. (*15*)		1	MR and skeletal abnormalities	oligo	44,290, 2,600	30-35 kb, 1 Mb	FISH
16	Decipher (*16*)		19	Syndromic phenotype including MR				
17	Koolen *et al*. (*17*)	386	29	MR with/or without congenital malformations and/or dysmorphism	BAC	32,447	100 kb	FISH, MLPA
	Total	**1349**	148					

(* checklist scoring based on de Vries *et al*, (*18*))

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