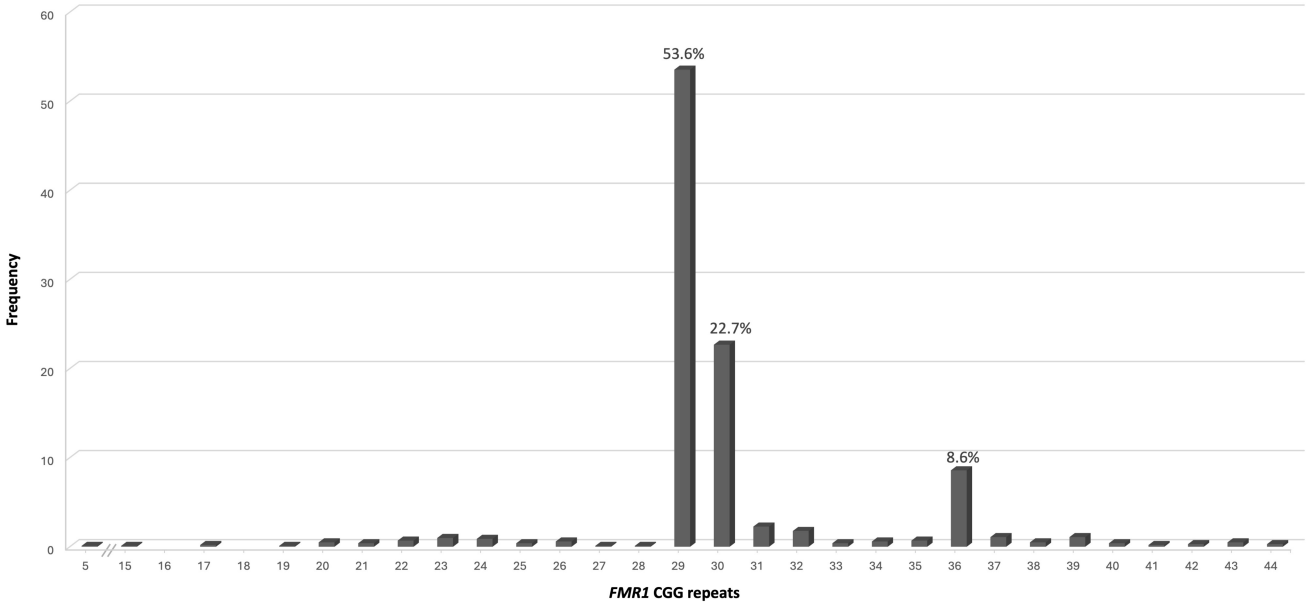


**Figure S1.** Distribution of normal CGG repeats in the *FMR1* gene among 1,014 male patients, which includes 93 males from a cohort of individuals with intellectual disability from the Child psychiatric clinic/Northern Thailand, and 921 males from a cohort of children undergoing routine fragile X syndrome testing at Songklanagarind Hospital, representing all regions of Thailand. The CGG repeat numbers were determined based on the results from fluorescent PCR followed by capillary electrophoresis. The most common alleles are 29 and 30 CGG repeats, followed by 36 CGG repeats.

**Table S1.** Full mutation expansions and AGG interruptions on *FMR1* maternal transmission

| Maternal repeat size | No. AGGs | Number of children with full mutation expansion |
|---|---|---|
| 70-79 | 0 AGG | 3[b] |
| | 1 AGG | 1 |
| 80-89 | 0 AGG | 4 |
| | 1 AGG | 2 |
| 90-99 | 0 AGG | 2 |
| | 1 AGG | 2 |
| 100-109 | 0 AGG | 7 |
| | 1 AGG | 2[b] |
| 110-119 | 0 AGG | 2 |
| | 1 AGG | 2 |
| 120-129 | 0 AGG | - |
| | 1 AGG | - |
| 130-139 | 0 AGG | - |
| | 1 AGG | - |
| 140-149 | 0 AGG | 1[a] |
| | 1 AGG | - |
| 150-199 | 0 AGG | 4[b] |
| | 1 AGG | - |
| >200 | 0 AGG | 2[b] |
| | 1 AGG | - |
| **Total** | | **34[c]** |

Please note that, in cases of patients with the full mutation in this cohort, no mother was found to carry alleles with < 70 CGG repeats.

Dash (-) indicates no mother carrier CGG repeats in this range.

[a] A mother carrier has 37/~140/>200 CGG repeats and has one child with a full mutation and another child with a mosaic premutation (~101 bp)/313 bp deletion [1].

[b] Mother in this group had more than one child with a full mutation.

[c] Among 34 patients with full mutations whose mothers were tested for AGG interruptions, TP-PCR analysis revealed 25 full mutation transmissions from maternal alleles with no AGG interruptions and nine transmissions from premutation alleles with one AGG interruption.

**Table S2.** Frequency of fragile X syndrome in high-risk groups worldwide using molecular testing (sample sizes ranging from 100 to >1000).

| Location | Subjects | No. of patients | Methods | FXS frequency (FM and mosaic FM) | Reference |
|---|---|---|---|---|---|
| **Asia** | | | | | |
| **Southeast Asia** | | | | | |
| Indonesia | Patients with DD | 262 | PCR with gel<br>SB | 1.5% (4/262)<br>M: 1.9% (4/206) | Faradz et al. 1999 [2] |
| Indonesia | Patient with ID, ASD, FHx of FXS | 176<br>M = 92<br>F = 84 | PCR with gel or CE<br>TP-PCR<br>SB | 4.0% (7/176)<br>M: 1.1% (1/92)<br>F: 7.1% (6/84) | Winarni et al. 2012 [3] |
| Malaysia | Patients with DD | 2,184<br>M = 2,057<br>F = 127 | PCR with gel or CE<br>MS-PCR<br>FragilEase kit | 3.5% (76/2,184)<br>M: 3.5% (73/2,057)<br>F: 2.4% (3/127) | Ali et al. 2017 [4] |
| Singapore | Males with LD | M = 255 | PCR with gel<br>SB | M: 2.4% (6/255) | Tan et al. 2000 [5] |
| Thailand | Patients with DD, ID, ASD | 1,480<br>M = 1,390<br>F = 90 | PCR with gel or CE<br>MS-PCR<br>TP-PCR<br>msTP-PCR<br>SB | 7% (103/1,480)<br>M: 7.2% (100/1,390)<br>F: 3.3% (3/90) | This study |
| **East Asia** | | | | | |
| China | Patients with ID | 1,127 | PCR with gel<br>SB | 2.8% (32/1,127)[a] | Zhong et al. 1999 [6] |
| China | Patients with ID | 324<br>M = 243<br>F = 81 | PCR with gel<br>SB | 0.6% (2/324)<br>M: 0.4% (1/243)<br>F: 1.2% (1/81) | Pang et al. 1999 [7] |
| China | Patients with ID, DD | 540<br>M = 453<br>F = 87 | PCR with gel<br>TP-PCR | 0.9% (5/540)<br>M: 1.1% (5/453)<br>F: 0% (0/87) | Chen et al. 2015 [8] |
| Japan | Patients with ID | 425<br>M = 296<br>F = 129 | PCR with gel<br>SB | 2.1% (9/425)<br>M: 2.4% (7/296)<br>F: 1.6% (2/129) | Hofstee et al. 1994 [9] |
| Japan | Males with ID | M = 256 | PCR with gel<br>SB | M: 0.8% (2/256) | Nanba et al. 1995 [10] |
| Japan | Patients with ASD | 109<br>M = 102<br>F = 7 | PCR with CE<br>SB | 0% (0/109)[a] | Otsuka et al. 2010 [11] |
| Korea | Patients with ID, LD, ASD | 101<br>M = 78<br>F = 23 | PCR with gel<br>SB | 1% (1/101)<br>M: 1.3% (1/78)<br>F: 0% (0/23) | Kwon et al. 2001 [12] |
| Taiwan | Patients with ID | 471<br>M = 311<br>F = 160 | PCR with gel<br>SB | 2.1% (10/471)<br>M: 2.9% (9/311)<br>F: 0.6% (1/160) | Tzeng et al. 2001 [13] |
| Taiwan | Patients with ID | 217<br>M = 148<br>F = 69 | PCR with gel<br>SB | 1.4% (3/217)<br>M: 1.4% (2/148)<br>F: 1.4% (1/69) | Yen et al. 2008 [14] |
| **South Asia** | | | | | |
| India | Patients with ID | 130<br>M = 93<br>F = 37 | PCR with gel<br>SB | 7.7% (10/130)<br>M: 9.7% (9/93)<br>F: 2.7% (1/37) | Sharma et al. 2001 [15] |

**Table S2.** (continued)

| Location | Subjects | No. of patients | Methods | FXS frequency (FM and mosaic FM) | Reference |
|---|---|---|---|---|---|
| India | Patients with ID | 146<br>M = 118<br>F = 28 | PCR with gel<br>SB | 2.1% (3/146)<br>M: 2.5% (3/118)<br>F: 0% (0/28) | Pandey et al. 2002 [16] |
| Pakistan | Patients with ID | 333<br>M = 229<br>F = 104 | PCR with gel<br>MS-PCR<br>SB | 4.8% (16/333)<br>M: 6.6% (15/229)<br>F: 1% (1/104) | Fatima et al. 2014 [17] |
| Pakistan | Patients with ID | 395<br>M = 287<br>F = 108 | PCR with gel<br>MS-PCR<br>SB | 3.3% (13/395)<br>M: 3.5% (10/287)<br>F: 2.8% (3/108) | Kanwal et al. 2015 [18] |
| Sri Lanka | Patients with ID, DD, ASD, ADHD | 850<br>M = 540<br>F = 310 | TP-PCR<br>MS-PCR<br>SB | 1.3% (11/850)<br>M: 2% (11/540)<br>F: 0% (0/310) | Chandrasekara et al. 2017 [19] |
| **Middle East** | | | | | |
| Iran | Patients with ID[b] | 508 (Proband)<br>M = 428<br>F = 80 | PCR with gel<br>SB | 6.3% (32/508)<br>M: 7.2% (31/428)<br>F: 1.3% (1/80) | Pouya et al. 2009 [20] |
| Kuwait | Males with ID | M = 182 | Cytogenetics<br>PCR with gel | M: 11% (20/182) | Bastaki et al. 2004 [21] |
| **Europe** | | | | | |
| Croatia | Patients with ID | 114<br>M = 73<br>F = 41 | PCR with gel<br>PCR-Chemi | 2.6% (3/114)<br>M: 2.7% (2/73)<br>F: 2.4% (1/41) | Hećimović et al. 2002 [22] |
| Estonia | Patients with ID | 516<br>M = 448<br>F = 68 | PCR with CE<br>SB | 2.7% (14/516)<br>M: 3.1% (14/448)<br>F: 0% (0/68) | Puusepp et al. 2008 [23] |
| France | Patients with ID | 574<br>M = 403<br>F = 171 | PCR<br>SB | 1.9% (11/574)<br>M: 2.5% (10/403)<br>F: 0.6% (1/171) | Gérard et al. 1997 [24] |
| France | Patients with ID | 20,816<br>M = 14,867<br>F = 5,949 | PCR<br>SB | 2.3% (477/20,816)<br>M: 2.8% (417/14,867)<br>F: 1% (60/5,949) | Biancalana et al. 2004 [25] |
| Greece | Patients with ID | 1,755<br>M = 1,426<br>F = 329 | PCR with CE<br>SB | 3.5% (61/1,755)<br>M: 4% (57/1,426)<br>F: 1.2% (4/329) | Sofocleous et al. 2008 [26] |
| Italy | Patients with ID, DD, ASD | 2,832<br>2,750 patients with ID, DD<br>82 patients with ASD | PCR with CE<br>TP-PCR<br>SB | 2.9% (82/2,832)[a]<br>ID, DD: 2.9% (81/2,750)<br>ASD: 1.2% (1/82) | Esposito et al. 2013 [27] |
| Latvia | Males with ID | M = 374 | PCR with CE<br>SB | M: 2.7% (10/374) | Daneberga et al. 2011 [28] |
| Netherland | Patients with ID | 236<br>M = 197<br>F = 39 | PCR with gel<br>SB | 4.2% (10/236)[a] | van den Ouweland et al. 1994 [29] |
| Netherland | Patients with ID | 1,531<br>M = 870<br>F = 661 | PCR with gel<br>SB | 0.7% (11/1,531)<br>M: 1% (9/870)<br>F: 0.3% (2/661) | de Vries et al. 1997 [30] |
| Spain | Patients with ID | 222<br>M = 182<br>F = 40 | SB | 5% (11/222)<br>M: 6% (11/182)<br>F: 0% (0/40) | Milà et al. 1997 [31] |

**Table S2.** (continued)

| Location | Subjects | No. of patients | Methods | FXS frequency (FM and mosaic FM) | Reference |
|---|---|---|---|---|---|
| Spain | Subjects with ID | 392<br>M = 277<br>F = 115 | PCR with gel<br>SB<br>Sequencing | 5.6% (22/392)<br>M: 6.5% (18/277)<br>F: 3.5% (4/115) | Millán et al. 1999 [32] |
| Turkey | Patients with ID, DD | 179<br>M = 166<br>F = 13 | PCR with gel<br>SB | 2.8% (5/179)<br>M: 3% (5/166)<br>F: 0% (0/13) | Tunçbilek et al. 1999 [33] |
| UK | Patients with LD | 154<br>M = 103<br>F = 51 | Cytogenetics<br>SB | 2.6% (4/154)<br>M: 3.9% (4/103)<br>F: 0% (0/51) | Slaney et al. 1995 [34] |
| UK | Males with LD | M = 3,732 | PCR<br>SB | M: 0.5% (20/3,732) | Youings et al. 2000 [35] |
| **North America** | | | | | |
| USA (Georgia) | Children in special education classes | 3,532<br>M = 2,471<br>F =1,061 | PCR<br>SB | 0.2% (7/3,532)<br>M: 0.3% (7/2,471)<br>F: 0% (0/1061) | Crawford et al. 2002 [36] |
| USA | Patients with ASD | 316 | PCR<br>SB | 1.9% (6/316)[a] | Reddy et al. 2005 [37] |
| USA | Males with mental dysfunction | M = 59,707 | PCR with gel or CE<br>SB | M: 1.4% (862/59,707) | Strom et al. 2007 [38] |
| USA | Patients with DD, ASD | 599<br>M = 486<br>F = 113 | PCR with CE<br>TP-PCR | 0.7% (4/599)<br>M: 0.6% (3/486)<br>F: 0.9% (1/113) | Tassone et al. 2013 [39] |
| **South America** | | | | | |
| Chile | Patients with ID and clinical features of FXS | 2,202 | PCR with gel or CE<br>SB | 8.2 % (181/2,202)[a] | Santa María L et al. 2016 [40] |
| Ecuador | Males with ID, ASD, language delay, FHx of FXS | M = 247 | PCR with gel | M: 8.9% (22/247) | Pozo-Palacios J et al. 2021 [41] |
| **Australia** | | | | | |
| Australia (Tasmania) | Patients with special educational needs (ID, LD, ASD, ADHD) | 1,248 | PCR with gel<br>SB | 0% (0/1,248)[a] | Mitchell et al. 2004 [42] |
| **Africa** | | | | | |
| Egypt | Males with ID, LD | M = 400 | PCR with gel | M: 4.8% (19/400) | Meguid et al. 2007 [43] |
| South Africa | Males with ID | 148 | SB | M: 6.1% (9/148) | Goldman et al. 1998 [44] |
| South Africa | Patients with ID | 2,239<br>M = 1,961<br>F = 278 | PCR with CE<br>SB | 5.7 % (128/2,239)<br>M: 6% (117/1,961)<br>F: 4% (11/278) | Essop et al. 2013 [45] |
| **Systematic review/ meta-analysis** | | | | | |
| Systematic review and meta-analysis | Patients with ID[c] | 7,475 | PCR<br>SB | 2.4% (178/7,475)[a] | Hunter et al. 2014 [46] |

ID, intellectual disability; ASD, autism spectrum disorder; DD, developmental delay; LD, learning disability; MS-PCR, methylation-specific PCR; PCR with gel, PCR followed by gel electrophoresis; PCR with CE, PCR followed

by capillary electrophoresis; PCR-Chemi, PCR followed by chemiluminescent detection; TP-PCR, triplet repeat-primed PCR; SB, Southern blot analysis; M, male; F, female; FM, full mutation (> 200 CGG repeats)

[a] Based on original research, the data does not provide frequencies separated by male and female; [b] Consanguineous and non-consanguineous parents; [c] Searching primary publication in PubMed, Embase, and the Cochrane library

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