

No.	Name	Gender	Age	Relationship to Fragile X	Wider Family links to Fragile X	Preconception Screening	Prenatal Screening	Newborn Screening
1	Malcolm	M	56	Parent of 23 year old son with FXS and unaffected daughter (donor conception)	None known.	✓✓	✓✓	✓✓
2	Judy	F	51	Parent of 23 year old son with FXS and unaffected daughter (donor conception). Wife of Malcolm	Cousin has FXS. Father has premutation.	✓	✓✓	XX
3	Mary	F	75	Spouse has FXTAS	Brother has FXS. Step-granddaughter with FXS	✓✓	XX	✓✓
4	Sue	F	74	Has FXTAS	Sister has FXTAS. Has two grandsons with FXS.	✓✓	X	✓✓
5	Ali	F	48	Premutation carrier, 2 unaffected daughters	One nephew and two nieces with FXS.	✓	XX	✓
6	Elisabete	F	31	Mother of 3 year old boy with FXS and unaffected daughter (prenatal testing)	None known.	✓	✓✓	XX
7	Emma	F	34	Mother of 14 year old boy with FXS and unaffected daughter (PGD)	Sister premutation carrier.	✓✓	✓✓	XX
8	Luca	M	17	Has FXS	Grandfather has FXTAS.	✓✓	XX	✓✓
9	Chiara	F	43	Mother of Luca (with FXS). Also has unaffected son (no prenatal testing)	Father has FXTAS. First cousin once removed has FXS.	✓	XX	✓✓
10	Joanne	F	47	Mother of two daughters with FXS (15, 12)	Sister has son with FXS.	✓✓	XX	✓
11	Valerie	F	66	Mother of Son (33) and daughter (29) both with FXS (no prenatal testing)	Cousin with FXTAS and daughter with FXS.	✓✓	XX	✓
12	Jane	F	58	Mother of son (25) and daughter (23) both with FXS. Also unaffected son (20) and daughter (19)(no prenatal testing)	Grandson (son of daughter, 23, with FXS) has FXS. Sister has daughter with FXS. First cousin once removed has son with FXS.	✓	X	✓✓

13	Sally-Anne	F	47	Mother of two daughters (15 and 11) with FXS. (no prenatal testing)	Father has FXTAS. Cousin has son with FXS.	✓✓	XX	✓✓
14	Zoe	F	40	Premutation carrier with carrier son (16) and unaffected daughter (12)	Two brothers with FXS.	✓	XX	✓✓
15	Holly	F	26	Has FXS.	Sister has FXS. Two male cousins have FXS.	✓✓	—	✓✓
16	Rachel	F	38	Mother of boy (10) with FXS and unaffected boy (2). (no prenatal testing)	Father had FXTAS (deceased). Aunt and uncle both premutation carriers. Six cousins all premutation carriers. Two cousins have sons with FXS and a third cousin currently pregnant with a boy with FXS.	✓	XX	✓
17	Tony	M	65	Grandparent of boy (7) with FXS	None known.	✓✓	—	✓✓
18	Moira	F	60	Grandparent of boy (10) with FXS	Both daughters premutation carriers.	✓	XX	✓
19	Kathryn	F	37	Mother of son (4)with FXS (undergoing PGD)	Sister has daughter with FXS. Cousin has son with FXS	✓✓	✓✓	✓✓

Table 1: Participant pseudonym, gender, age, relationship to fragile X syndrome and attitudes towards screening.

(✓✓= support for screening, ✓= ambivalent support for screening, XX= non-support for screening, X= ambivalent non-support for screening, -- = neutral/unable to take a view)