

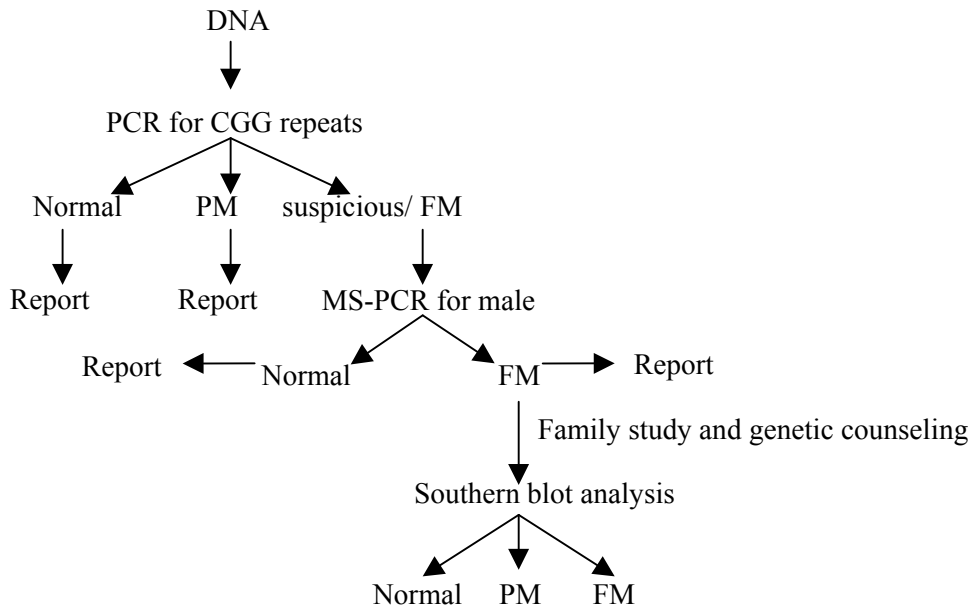
4. Conclusion

The study has shown that MS-PCR is an accurate method for diagnosis of FXS, comparable to the current gold standard methods. MS-PCR can determine methylation status and an approximate number for CGG repeats, as in Southern blot analysis. In addition, because it is a PCR-based method, it requires only a small amount of DNA and is less time consuming, which can be important in some circumstances such as prenatal diagnosis and analysis of DNA extracted from small blood or tissue samples. Also in this study, we performed prenatal diagnosis using MS-PCR, which has not previously been reported, and based on this result we can report that it is possible that MS-PCR might be able to replace Southern blot analysis for a prenatal diagnosis in known FXS families.

Finally, we propose a new guideline for diagnosis of FXS (Fig. 24). Integration of MS-PCR into the current practice might reduce the number of cases which require Southern blot analysis, and thus also reduce the cost of diagnosis. In case of prenatal diagnosis, PCR for *SRY* gene to determine fetal sex is included (Charalsawad, et al. 2000).

Guideline for fragile X syndrome DNA diagnosis

A. Male DNA sample



B. Female DNA sample

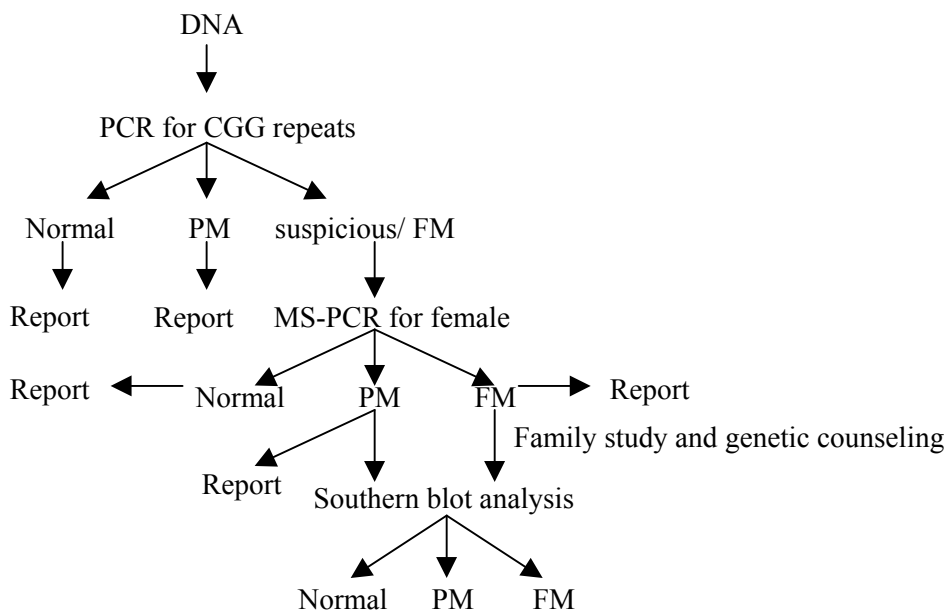


Fig. 24 Schematic illustration of the guideline for diagnosis of FXS. Guideline for a male DNA sample (A) and a female DNA sample (B).