**58**

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# FAMILIAL PELVIC FLOOR DISORDER CASES ARE MORE LIKELY TO HAVE A MIXED PHENOTYPE

Hypothesis / aims of study

Pelvic floor disorders (PFDs) include a variety of continence and organ laxity defects; many women with PFDs have more than one defect. Understanding the clustering of PFDs within families is important because it may suggest underlying risk factors that may be environmental, genetic or both. The aim of this study was to describe clusters of familial cases with PFDs as well as clinical characteristics of familial cases.

Study design, materials and methods

Women evaluated and treated for PFDs were recruited as part of a larger genetic study; here we define familial cases as those with at least bothersome symptoms for a PFD (pelvic organ prolapse- POP, stress urinary incontinence-SUI and overactive bladder- OAB) and who had a first degree relative with at least bothersome symptoms of the same pelvic floor defect. Probands and their relatives were clinically phenotyped using standardized symptom questions (PFDI), demographic data, examination including clinical markers such as striae or joint mobility, and review of treatment records, if any. “Bothersome” was defined as bothered “moderately” or “quite a bit” on the PFDI, or any treatment.

Results

In our resource, we identified 126 familial POP cases, 183 familial SUI cases and 101 familial OAB cases. All familial cases were more likely to have bothersome symptoms in more than one PFD. Among familial POP cases, at least bothersome SUI (73%), OAB (54%), and combination of all three disorders (49.2%) were common. Among familial SUI cases, at least bothersome OAB (59%), POP (57.4%), and combinations of all disorders (38.3%) were common. Among familial OAB cases, at least bothersome SUI (89.1%), POP (64.4%), and combinations of all three disorders (58.4%) were common. Table 1 shows clinical characteristics of these familial cases with results being similar across the three groups studied.

| Characteristic | Familial POP | Familial SUI | Familial OAB |
|---|---|---|---|
| Number | 126 | 183 | 101 |
| Mean age at diagnosis (SD) | 49.7 (14.3) (n=83) | 46.2 (13.4) (n=95) | 49.2 (15.6) (n=48) |
| Mean BMI (SD) | 27.0 (5.7) (n=117) | 27.2 (5.7) (n=156) | 27.5 (5.4) (n=81) |
| Mean parity (SD) | 4.1 (2.3) (n=117) | 3.7 (2.3) (n=159) | 3.9 (2.3)(n=90) |
| Mean highest birth wt(g)(SD) | 3,721 (495) (n=112) | 3,703 (568) (n=151) | 3,690 (510)(n=83) |
| Percent with joint mobility | 42.0% (n=112) | 38.6% (n=140) | 33.3% (n=75) |
| Percent with striae | 60.2% (n=123) | 62.4% (n=170) | 60.6% (n=94) |

Interpretation of results

It is unknown whether pelvic floor disorders are multiple manisfestations of a single disorder, or discrete conditions that share common risk factors but not an underlying etiology. In families with multiple cases of pelvic floor disorders, women are more likely to have a mixed phenotype suggesting that these defects share common underlying etiologic factors.

Concluding message

Familial cases of POP, SUI and OAB are more likely to have more than one pelvic floor defect. It is likely that underlying genetic factors contribute to more than one pelvic floor defect.

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| ***Is this a clinical trial?*** | **No** |
| ***What were the subjects in the study?*** | **HUMAN** |
| ***Was this study approved by an ethics committee?*** | **Yes** |
| ***Specify Name of Ethics Committee*** | **University of Utah Institutional Review Board** |
| ***Was the Declaration of Helsinki followed?*** | **Yes** |
| ***Was informed consent obtained from the patients?*** | **Yes** |