|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Table 2.** Clinical features of biliary tract disease in individuals with Rett syndrome and/or *MECP2* mutations from the Natural History of Rett Syndrome Study (n=28) | | | | | |
|  | Biliary Tract Disease Present | | Biliary Tract Disease Absent | |  |
| Variable | Number of Abnormal Responses | Number of Respondents | Number of Abnormal Responses | Number of Respondents | P-value |
| Age (y) | 25 (21, 34) | 12 | 21 (11, 24) | 16 | <0.05 |
| Gender (female) | -- | 12 | -- | 16 | NS |
| Diagnostic criteria\* |  | 11 |  | 12 | NS |
| Clinical +, mutation – | 2 |  | 0 |  |  |
| Clinical +, mutation + | 8 |  | 12 |  |  |
| Clinical –, mutation + | 1 |  | 0 |  |  |
| Clinical severity of mutation\*\* |  | 11 |  | 15 | NS |
| Mild | 1 |  | 3 |  |  |
| Moderate | 7 |  | 10 |  |  |
| Severe | 3 |  | 2 |  |  |
| Positive family history | 7 | 11 | 4 | 15 | <0.06 |
| Symptoms |  |  |  |  |  |
| Abdominal pain | 7 | 7 | 5 | 7 | NS |
| Irritability | 4 | 8 | 7 | 10 | NS |
| Vomiting | 4 | 7 | 2 | 5 | NS |
| Fever | 1 | 6 | 2 | 7 | NS |
| Duration of symptoms (mo) | 21 (3, 72) | 6 | ― | ― |  |
| Diagnostic tests |  |  |  |  |  |
| Abdominal ultrasound | 8 | 11 | 0 | 11 | <0.001 |
| Abdominal CT scan | 3 | 4 | 0 | 4 | NS |
| CCK-HIDA scan | 5 | 5 | 0 | 5 | <0.01 |
| Surgery | 10 | 12 | 0 | 16 | <0.001 |
| \* Neul J, 2010 (see reference 2)  \*\*Culdepah V, 2014 (see reference 6) | | | | | |