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| **Table 1.** Clinical features of biliary tract disease in 46 girls and women with Rett syndrome identified by medical record review |
| Variable | Value\* | Number of Individuals |
| Caucasian (%) | 90 | 28 |
| Age (y) at first symptoms  | 21 (14, 24) | 37 |
| Presenting symptoms |  |  |
|  Abdominal pain (%) | 94 | 36 |
|  Irritability (%) | 88 | 18 |
|  Weight loss (%) | 64 | 11 |
|  Vomiting (%) | 52 | 25 |
|  Fever (%) | 33 | 14 |
| Duration (mo) of symptoms  | 2.5 (1, 12) | 30 |
| Family history positive (%) | 65 | 23 |
| Growth measurements |  |  |
|  Height (z-score)  | -2.5 (-3.2, -1.5) | 19 |
|  Weight (z-score)  | -3.0 (-4.2, -0.3) | 34 |
|  Body mass index (z-score)  | -0.4 (-1.3, 1.1) | 19 |
| Physical exam |  |  |
|  Right upper quadrant tenderness (%) | 24 | 25 |
|  Abdominal distention (%) | 24 | 21 |
| Laboratory Tests |  |  |
|  AST (u/l) | 29 (17, 40) | 25 |
|  ALT (u/l) | 26 (19, 41) | 22 |
|  Alkaline phosphatase (u/l) | 108 (75, 180) | 25 |
|  Total bilirubin (mg/dl) | 0.4 (0.3, 0.6) | 21 |
| Abdominal ultrasound |  |  |
|  Gallstones (%) | 70 | 40 |
|  Dilated duct (%) | 15 | 26 |
| CT scan |  |  |
|  Gallstones (%) | 31 | 13 |
| CCK-HIDA scan |  |  |
|  Delayed emptying (%) | 90 | 21 |
| Age (y) at diagnosis  | 21 (15, 26) | 39 |
| Surgery (%) | 85 | 46 |
| Procedure  |  |  |
|  Laparoscopy (%) | 81 | 37 |
| Age (y) at surgery | 21 (15, 26) | 38 |
| Pathology |  |  |
|  Cholecystitis (%) | 77 | 31 |
|  Cholelithiasis (%) | 65 | 31 |
| \*Values expressed as median, first and third quartiles or proportion (%) where indicated |