**Supplementary Table 3. Diseases associated with breast cancer susceptibility genes**

|  |  |
| --- | --- |
| **Disease** | **Breast Cancer Susceptibility Genes** |
| *ATM* | *BARD1* | *BRCA1* | *BRCA2* | *CDH1* | *CHEK2* | *NF1* | *PALB2* | *PTEN* | *RECQL* | *STK11* | *TP53* |
| Adrenocortical Carcinoma |   |   |   |   |   |   |   |   |   |   |   | 1 |
| Autism |   |   |   |   |   |   |   |   | 1 |   |   |   |
| BCD Syndrome\* |   |   |   |   | 1 |   |   |   |   |   |   |   |
| Bone Dysplasia |   |   |   |   |   |   | 1 |   |   |   |   |   |
| Brain Tumor |   |   |   |  |   |   | 1 |   | 1 |   |   | 1 |
| Breast Cancer | 1 | 1 | 1 | 1 | 1 | 1 | 1 | 1 | 1 | 1 | 1 | 1 |
| Cafe-Au-Lait Spots |   |   |   |   |   |   | 1 |   |   |   |   |   |
| Cerebrovascular Malformation |   |   |   |   |   |   |   |   | 1 |   |   |   |
| Cervical Cancer |   |   |   |   |   |   |   |   |   |   | 1 |   |
| Colorectal Cancer | 1 |   |   |   |   | 1 |   |   | 1 |   | 1 | 1 |
| Endometrial Cancer |   |   |   |   |   |   |   |   | 1 |   | 1 |   |
| Facial Papules |   |   |   |   |   |   |   |   | 1 |   |   |   |
| Gastric Cancer | 1 |   |   |   | 1 | 1 |   | 1  |   |   | 1 |   |
| GI Hamartomatous Polyps |   |   |   |   |   |   |   |   | 1 |   | 1 |   |
| GIST |   |   |   |   |   |   | 1 |   |   |   |   |   |
| Hepatobiliary Cancer |   |   |   |   |   |   |   |   |   |   | 1 | 1 |
| Intellectual Disability |   |   |   |   |   |   | 1 |   |   |   |   |   |
| Iris Hamartoma |   |   |   |   |   |   | 1 |   |   |   |   |   |
| Kidney Cancer |   |   |   |   |   | 1 |   |   | 1 |   |   |   |
| Leukemia |   |   |   |   |   |   | 1 |   |   |   |   |   |
| Lipoma |   |   |   |   |   |   |   |   | 1 |   |   |   |
| Lung Cancer |   |   |   |   |   |   |   |   |   |   | 1 |   |
| Macrocephaly |   |   |   |   |   |   |   |   | 1 |   |   |   |
| Melanoma |   |   |   | 1 |   |   |   |   | 1 |   |   |   |
| Neurofibroma |   |   |   |   |   |   | 1 |   |   |   |   |   |
| Non-Epithelial Ovarian Tumor |  |  |  |  |  |  |  |  |  |  | 1 |  |
| Oral Mucosal Papillomatosis |   |   |   |   |   |   |   |   | 1 |   |   |   |
| Ovarian Cancer |   |   | 1 | 1 |   |   |   | 1 |   |   |   |   |
| Ovarian SCST |   |   |   |   |   |   |   |   |   |   | 1 |   |
| Osteosarcoma |  |  |  |  |  |  |  |  |  |  |  | 1 |
| Pancreatic Cancer | 1 |   | 1 | 1 |   |   |   | 1 |   |   | 1 | 1 |
| Paraganglioma |   |   |   |   |   |   | 1 |   |   |   |   |   |
| Pheochromocytoma |   |   |   |   |   |   | 1 |   |   |   |   |   |
| Prostate Cancer | 1 |   | 1 | 1 |  | 1 |   | 1 |  |   |   |   |
| Pulmonary Stenosis |   |   |   |   |   |   | 1 |   |   |   |   |   |
| Sarcoma |   |   |   |   |   | 1 | 1 |   |   |   |   | 1 |
| Skin (Benign)\*\* |   |   |   |   |   |   | 1 |   | 1 |   | 1 |   |
| Small Intestine Cancer |   |   |   |   |   |   |   |   |   |   | 1 |   |
| Testicular SCST |   |   |   |   |   |   |   |   |   |   | 1 |   |
| Thyroid (Benign)\*\* |   |   |   |   |   |   |   |   | 1 |   |   |   |
| Thyroid Cancer |   |   |   |   |   | 1 |   |   | 1 |   |   |   |
| Uterine Fibroid |   |   |   |   |   |   |   |   | 1 |   |   |   |

Abbreviations: GI, gastrointestinal; BCD, blepharocheilodontic; SCST, sex cord-stromal tumor; GIST, gastrointestinal stromal tumor

Note: The number ‘1’ indicates that the gene was associated with the disease/cancer in the resource. Blank space indicates association was not found in the resource.

\* BCD syndrome consists of: Facial Dysmorphism, Hypertelorism, Imperforate anus, Distichiasis, Clinodactyly, Hypoplastic nails, Choanal atresia, Cleft palate, benign teeth disorder.

\*\*Benign: This may mean a verity of benign diseases related to the organ and have been elaborated in Supplement Table 1