

Additional file 2. Biological functions related to the list of 109 genes and their involvement in the phenotype in the current patient.

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| Outer dyneine arm assembly |
| Inner dyneine arm assembly |
| Brain development |
| Gland development |
| Gonad development |
| Head development |
| Development of the anatomical structure |
| Development of the trachea |
| Development of primary sexual characteristics |
| Forebrain development |
| Development of the heart |
| Development of the endocrine system |
| Development of the central nervous system |
| Development of the respiratory system |
| Liver development |
| Determination of left/right asymmetry in the nervous system |
| Determination of the male sex |
| Primary sex determination |
| Sertoli cell differentiation |
| Sexual differentiation |
| Axoneme assembly |
| Cilium assembly |
| Assembly of the axonemal dynein complex |
| Cell motility |
| Cilium-dependent cell motility and movement of the cilium |
| Microtubule-based movement |
| Movement of the epithelial cilium involved in the movement of extracellular fluid |
| Organization of cellular components |
| Organization of the cytoskeleton |
| Organization of the microtubule cytoskeleton |
| Microtubule-based process |
| Various development process |
| Cellular chemotaxis |
| Negative regulation of neural precursor cell proliferation. |
| Positive regulation of male gonad development |
| Reproduction and reproductive process |