

Additional file 3. Genotype-phenotype association of the list of 109 genes in the current patient.

Miscellaneous phenotypic abnormality
Functional abnormality of the male internal genitalia
Morphological abnormality of the gastrointestinal tract
Head abnormality
Abnormality of the physiology of the reproductive system
Abnormality of the female external genitalia
Abnormality of the male external genitalia
Abnormality of the male internal genitalia
Abnormality of the abdominal organs.
Abnormality of prenatal development or birth
Epididymal abnormality
Ovarian abnormality
Abnormality of the genital system
Abnormality of the genitourinary system
Immune system abnormality
Abnormality of the respiratory system.
Gastrointestinal tract abnormality
Upper respiratory tract abnormality
Absence of secondary sexual characteristics
Azoospermia
Internal and external dynein arms absent
Stomach cancer
Ovarian band
Dextrocardia
Ciliary dyskinesia
Gonadal dysgenesis
Testicular dysgenesis
Decreased serum testosterone concentration
Decreased fertility in females.
Cone-rod dystrophy
Peribronchovascular interstitial thickening
Sterility
Female external genitalia in an individual with 46,XY karyotype
Clitoral hypertrophy
Hypergonadotropic hypogonadism
Hypogonadotropic hypogonadism
Vaginal hypoplasia
Ovarian hypoplasia
Feminine infertility
Male infertility

Primary gonadal failure
Suprarrenal insufficiency
Sex inversion
Intestinal malrotation
Abnormal morphology of the vagina
Abnormal morphology of the inferior vena cava
Abnormal morphology of fingers
Abnormal morphology of the female internal genitalia
Abnormal morphology of the vaginal lips
Abnormal morphology of the respiratory motile cilium
Abnormal morphology of the heart
Abnormal bowel morphology
Abnormal lung morphology
Polycystic ovaries
Sparse hair
Male pseudohermaphroditism
Delayed puberty
Puberty and gonadal disorders
Delayed speech and language development
Neurodevelopmental delay
Astrocytoma Risk
Ovarian Gonadoblastoma Risk
Testicular Gonadoblastoma Risk
Ovarian Neoplasia Risk
Risk of Genitourinary Tract Neoplasia
Risk of gonadal neoplasia
Risk of Renal Neoplasia
Risk of testicular neoplasia
Gonadoblastoma Risk
Pulmonary ambiguous situation
Situs inversus totalis
Absent or hypoplastic testicles
Variation of circulating gonadotropin level
Variation in circulating follicle-stimulating hormone levels
Variation in circulating luteinizing hormone levels
Serum estradiol variations
Variations in the measure of sex hormones
Sparse armpit hair
Sparse pubic hair