Swyer Syndrome

Other Names:
46, XY CGD; 46, XY complete gonadal dysgenesis; 46, XY pure gonadal dysgenesis; See More

Categories:
Blood Diseases; Congenital and Genetic Diseases; Endocrine Diseases; See More

Summary

Swyer syndrome is a condition in which people with one X chromosome and one Y chromosome (normally present in males) have a female appearance. People with Swyer syndrome are typically raised as females, have a female gender identity, have typical female external genitalia, and have a normal uterus and Fallopian tubes. However in place of functional gonads (ovaries or testes), they have undeveloped, residual gonadal tissue called streak gonads. Streak gonads often become cancerous, so they are usually surgically removed as early as possible. Swyer syndrome may be caused by mutations in any of several genes. The inheritance pattern depends on the responsible gene.[1][2] In addition to removal of streak gonads, treatment may include hormone replacement therapy from puberty onward. While women with Swyer syndrome are infertile, they may become pregnant with the use of donated eggs.[3]

Last updated: 11/16/2015

Symptoms

This table lists symptoms that people with this disease may have. For most diseases, symptoms will vary from person to person. People with the same disease may not have all the symptoms listed. This information comes from a database called the Human Phenotype Ontology (HPO). The HPO collects information on symptoms that have been described in medical resources. The HPO is updated regularly. Use the HPO ID to access more in-depth information about a symptom.

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