## Our tests

improve the health of entire families.

**Identify** members of a family who carry the disease-causing mutation.

**Focus treatment** on relatives at risk.

**Eliminate** from surveillance relatives who are not at risk.

**Impact Genetics** performs molecular diagnostic tests for retinoblastoma families from across Canada, the United States and many other countries.

The **Impact Genetics** retinoblastoma test uses a series of different molecular assays to identify the unique *RB1* gene mutations or *MYCN* copy number change that initiate retinal tumors. Our tests identify the disease-causing error 96% of the time - **the highest reported pick-up rate worldwide.**

impact genetics inc.

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advancing genetic diagnostics

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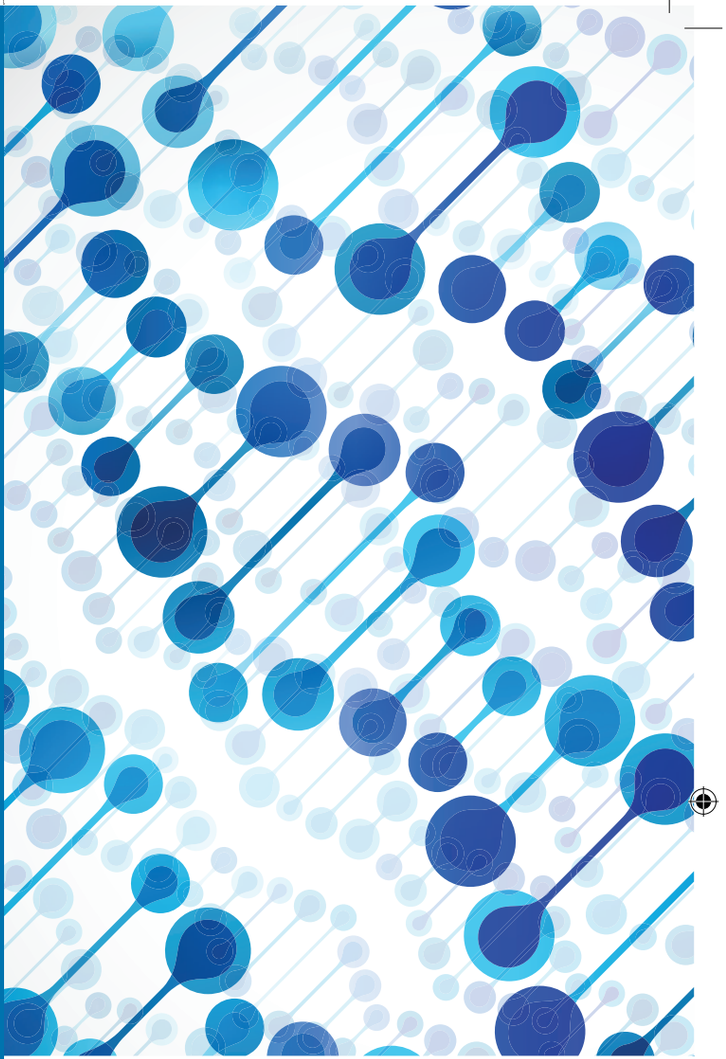
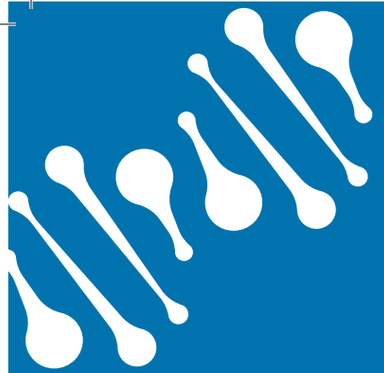


# Bilateral RETINOBLASTOMA

## Benefits of genetic testing



## What is Retinoblastoma?

Retinoblastoma *(Reh-tin-oh-blast-oma)* is a cancer of one or both eyes that occurs in young children. Retinoblastoma affects about 1 in 15,000 live births, and an estimated 9,000 children develop the cancer each year around the world. Retinoblastoma affects children of all races and both boys and girls.

The retinoblastoma tumor(s) originate in the retina, the light sensitive layer of the eye that enables the eye to see. When the tumors are present in one eye, it is referred to as unilateral retinoblastoma, and when it occurs in both eyes it is referred to as bilateral retinoblastoma. 60% of cases involve only one eye (unilateral) and the rest (40%) affect both eyes (bilateral). The majority (90%) of retinoblastoma patients have no family history of the disease.

The most common early sign of retinoblastoma is a white glow in the child's eye. This cancer is easy to diagnose, and treatment is very effective when tumors are found early.

## Genetics

Errors (mutations) in the *RB1* gene initiate retinal tumors. *RB1* mutations can be inherited from a parent (10%), but most occur spontaneously pre-conception or during the baby's early development (90%). In 2% of unilateral retinoblastoma, a retinal tumor is initiated by multiple copies of the *MYCN* gene (*RB1* gene is normal).

DNA from blood, tumor, amniocytes or other specimen types is tested in our licensed lab.

At **Impact Genetics**, we detect 96% of *RB1* mutations.

# BENEFITS OF GENETIC TESTING

# Bilateral Retinoblastoma *(cancer in both eyes)*

## 1 Family

Knowing the disease causing genetic mutation in the patient makes it possible to look for the same mutation in family members. Surveillance and treatment of family members is improved if we know whether they carry the mutation.

| | without genetic testing | with genetic testing* | |
|---|---|---|---|
| Parents | · one retinal exam | 95% test negative | · one retinal exam for retinoma |
| | | 5% test positive | · one retinal exam for retinoma<br>· cancer surveillance<br>· changes risk for future offspring |
| Siblings | · multiple exams under anesthetic | 97% test negative | · NO exams required |
| | | 3% test positive | · multiple exams under anesthetic<br>· cancer surveillance |
| Offspring | · multiple exams under anesthetic | 50% test negative | · NO exams required |
| | | 50% test positive | · if tested in pregnancy, early delivery<br>· multiple exams under anesthetic<br>· cancer surveillance |

* Positive and negative test proportions apply to families where only one individual is affected with retinoblastoma. Data based on research by Dr. B. Gallie and Impact Genetics. Not to replace best judgement of medical practitioner.

## 2 Future cancer surveillance

If the genetic mutations of a tumor (retinoblastoma or second cancer) are known, a highly sensitive test may be used to monitor for spread of cancer.

impactgenetics.com