

## BC Newborn Screening Program

## Information Sheet

### Congenital Hypothyroidism (CH) – Endocrine Disorder

#### What is Congenital Hypothyroidism (CH)?

CH is a condition in which babies are born with a thyroid hormone deficiency. Thyroid hormone is important for the normal function of all the body's organs and for brain development.

#### What is its incidence?

Congenital Hypothyroidism is relatively common condition that affects about 1 in every 4,000 babies born in BC.

#### What causes the disease?

The most common causes of congenital hypothyroidism are partial or total failure of the thyroid gland to develop and defects in thyroid hormone synthesis. Congenital Hypothyroidism is a heterogeneous disorder and likely to have many different causes, both genetic and non-genetic.

#### What are the clinical features of the disease?

The majority of babies with Congenital Hypothyroidism appear normal at birth. Some may have jaundice, constipation, lethargy, hypotonia, and feeding problems. However, if they are not identified and treated quickly, they are at risk for mental retardation and failure to thrive.

#### How is the diagnosis confirmed?

Screening for Congenital Hypothyroidism is done by measuring thyroid stimulating hormone (TSH) levels in dried blood spots. The diagnosis is confirmed with a repeat measurement of TSH and free thyroxine using a blood sample. Further tests can

include thyroid scans and X-rays.

Diagnostic testing is arranged by specialists at BC Children's Hospital.

#### What is the treatment of the disease?

Thyroid hormone replacement in the form of thyroxine is an effective treatment to prevent the symptoms of hypothyroidism. Treatment is lifelong but is simple to administer and readily available. Specialists at BC Children's Hospital will coordinate treatment.

#### What is the outcome of treatment?

Infants who are identified early and treated appropriately have an excellent prognosis and grow up to be indistinguishable from other children the same age.

#### How can the disease be prevented?

Since there are many different reasons why CH can occur, some parents who have a child with congenital hypothyroidism can have an increased chance to have another child with the same condition while others do not.

#### Resources

<http://www.nlm.nih.gov/medlineplus/ency/article/001193.htm>

<http://www.geneclinics.org>

Revised November 2009