Tuberous Sclerosis Complex (TSC)

1 in 6000 babies are born with Tuberous Sclerosis Complex (TSC)

Genetics professionals such as medical geneticists and genetic counsellors can discuss conditions like TSC in more detail and answer any questions you may have about genetic results. To find a genetics clinic near you visit the Canadian Association of Genetic Counsellors website www.cagc-accg.ca.

There are many online resources for individuals and families coping with the diagnosis and management of TSC, some of which are listed below.

- Tuberous Sclerosis Canada: http://www.tscanada.ca/
- Tuberous Sclerosis Alliance: http://www.tsalliance.org
- TSC Canadian Clinics:
  - Ste. Justine Hospital, Montreal QC
  - BC Children’s Hospital, Vancouver BC
  - Toronto General Hospital, Toronto ON

The results of genetic testing can provide individuals and their families with important information by:
- Confirming a diagnosis, particularly when clinical findings are unclear
- Guiding treatment decisions, thereby improving outcome
- Providing important information about prognosis and future health concerns
- Clarifying risks to family members
- Empowering families to make family planning decisions

GENETIC TESTING CAN HELP

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GENETIC TESTING CAN HELP

The Field Of Genetics Is Always Evolving And So Are We Please Visit Our Website For A Current Test List

REFERENCES

RESOURCES

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Table 2. Major and minor features of TSC

<table>
<thead>
<tr>
<th>MAJOR FEATURES</th>
<th>MINOR FEATURES</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypomelanotic macules (&gt;3, at least 5-mm diameter)</td>
<td>Confetti skin lesions</td>
</tr>
<tr>
<td>Angiofibromas (&gt;3) or fibrous cephalic plaque</td>
<td>Dental enamel pits (&gt;3)</td>
</tr>
<tr>
<td>Vignal fibromas (&gt;2)</td>
<td>Intraoral fibromas (&gt;2)</td>
</tr>
<tr>
<td>Shagreen patch</td>
<td>Retinal achromatic patch</td>
</tr>
<tr>
<td>Multiple retinal hamartomas</td>
<td>Multiple renal cysts</td>
</tr>
<tr>
<td>Cortical dysplasia</td>
<td>Nonrenal hamartomas</td>
</tr>
<tr>
<td>Subependymal nodules</td>
<td>Cardiac rhabdomyoma</td>
</tr>
<tr>
<td>Subependymal giant cell astrocytoma</td>
<td>Lymphangiomyomatosis (LAM)*</td>
</tr>
<tr>
<td>Cardiac rhabdomyoma</td>
<td>Angiomyolipomas (&gt;2)*</td>
</tr>
<tr>
<td>Lymphangioleiomyomatosis (LAM)*</td>
<td>Angiomyolipomas (&gt;2)*</td>
</tr>
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</table>

* A combination of the two major clinical features (LAM and angiomyolipomas) without other features does not meet criteria for a definite diagnosis.
THE FACTS ABOUT TSC

WHAT YOU NEED TO KNOW

- Affects over 3500 Canadians.
- TSC causes tumours to develop in vital organs, such as the brain, eyes, heart, kidneys, skin and lungs.
- Most tumours are benign.
- When heart tumours are present, they may be seen on prenatal ultrasound, providing an early diagnosis of TSC.
- TSC is the leading genetic cause of autism and epilepsy.
- 90%-95% have seizures.
- Up to 60% are diagnosed with autism.
- TSC is 100% penetrant, meaning that if someone has a genetic mutation in TSC1 or TSC2, the individual will have clinical features of TSC. The severity and the age of onset of TSC features can vary from person to person and even within families.
- Early diagnosis is key to early intervention, which is important to treat tumors, control seizures, and improve learning and development.

GENETICS PLAYS AN IMPORTANT ROLE

- 85-90% of individuals will have a genetic change (or mutation) in genes TSC1 or TSC2.
- In the other 10-15% of cases, a negative genetic test does not mean that someone does not have TSC.
- Two thirds of cases result from a new mutation (first case in the family) and one third of the time, TSC is inherited from an affected parent.
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