

Guy's and St Thomas'

NHS Foundation Trust



Beckwith- Wiedemann Syndrome: tumour risk and surveillance

This leaflet has information about the risks and surveillance (monitoring) of tumours in children with Beckwith-Wiedemann Syndrome.

What is Beckwith-Wiedemann Syndrome (BWS)?

BWS is a condition that affects many parts of the body. About one in 15,000 people are born with BWS. It is known as an overgrowth syndrome, which means patients are larger than expected.

Some patients with BWS have:

- higher birth weight
- longer length on one side of the body (asymmetry)
- enlarged tongue
- unusual blood vessels
- openings in the abdominal (tummy) wall
- a crease in the earlobe or a pit behind the ear.

The physical signs and symptoms of BWS vary and few children have all of the characteristics.

Chromosomes and genes

People usually inherit 23 pairs of chromosomes, one of each pair from their mother and the other from their father.

Genes also come in pairs and lie along the chromosomes. Genes are the body's instructions for growth and development. Usually, both copies of a gene need to be 'switched on' when it is time for them to do their job.

For some genes only one copy of the gene is switched on depending which parent it has been inherited from. This process is called 'genomic imprinting'. Problems with genomic imprinting of genes on chromosome 11 are responsible for most cases of BWS.

What causes BWS?

About 50% (half) of cases occur due to errors in 'genomic imprinting' of one or more genes on chromosome 11. This alters how these genes do their usual job.

In about 20% (one in five) of cases, BWS occurs when two copies of chromosome 11 have come from the father and no copies have come from the mother.

Less commonly, BWS is caused by a 'mutation' (spelling mistake) in the CDKN1C gene or other changes involving the imprinted region on chromosome 11.

In about 85% of cases, BWS is not inherited. Please discuss questions of heredity with your genetics team.

What are the tumour risks?

The tumour risks depend on the underlying cause of BWS. All affected individuals have a chance of developing different tumours but it is difficult to estimate the risk for each type. Experience shows that most children with BWS will not develop a tumour at all.

The main tumour risk in childhood is for Wilms tumour (a kidney tumour). There are also increased risks of hepatoblastoma in the liver, neuroblastoma in the adrenal gland or elsewhere and rhabdomyosarcoma (in the muscle or connective tissue) amongst others.

Children with BWS are at the highest risk of developing tumours up to about eight years of age. The risk then falls over time. In adult life, the risk is similar to that of the general population.

Tumour surveillance (monitoring)

Monitoring cannot prevent tumours from developing, but may identify a tumour sooner so that treatment can begin earlier.

Surveillance is aimed mainly at the detection of Wilms tumours, as the other tumours are harder to detect.

The South East Thames Regional Genetics Service suggests the following surveillance, based on our team's experience and published reports:

- abdominal ultrasound scans every three months until the age of eight to look for Wilms and hepatoblastoma
- blood tests to check serum alpha-fetoprotein (AFP) levels every two to three months until the age of four.

Surveillance should be performed after detailed discussion with a clinical geneticist and paediatrician regarding the benefits and limitations of the tests.

Guidelines

In the UK, there are no national guidelines. We recognise that this is a complex issue, due to uncertainty about the benefits of monitoring.

What are the risks and limitations?

As with all surveillance, some tumours will not be detected due to limitations in screening methods or because of the tumour size and/or location. Monitoring could give you a 'false positive' result, suggesting a tumour is present when it is not, thus causing anxiety in the family and leading to further investigations.

Frequent scans and blood tests can be stressful for you and your child. Therefore, we encourage families to discuss the issues so that informed decisions can be made.

Seen in clinic by:

Notes:

Contact us

If you have any questions or concerns about the information in this leaflet, please contact the Clinical Genetics Department on 020 7188 1364.

Find out more about the service at www.guysandstthomas.nhs.uk/our-services/genetics/overview.aspx

For more information leaflets on conditions, procedures, treatments and services offered at our hospitals, please visit www.guysandstthomas.nhs.uk/leaflets

Pharmacy Medicines Helpline

If you have any questions or concerns about your medicines, please speak to the staff caring for you or call our helpline.

t: 020 7188 8748, 9am to 5pm, Monday to Friday

Your comments and concerns

For advice, support or to raise a concern, contact our Patient Advice and Liaison Service (PALS). To make a complaint, contact the complaints department.

t: 020 7188 8801 (PALS) **e:** pals@gstt.nhs.uk

t: 020 7188 3514 (complaints)

e: complaints2@gstt.nhs.uk

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