FACTSHEET #12
CANCER

Noonan Syndrome Association

People with Noonan syndrome have a small increased risk of several blood-related (haematological) cancers.

The genes involved in Noonan syndrome are part of the RAS/MAPK pathway, which controls cell growth.

Mutations in these genes which are present at birth and occurred in egg or sperm (germinal mutations) such as in Noonan Syndrome are not usually associated with cancer. However when they occur due to a genetic error in the body after birth (somatic mutations) they may lead to a local cancers.

Because somatic mutations of these genes may cause cancer they have been studied extensively by the pharmaceutical industry and there are many drugs in the pipeline which block the RAS/MAPK pathway and could potentially have application for treatment in Noonan syndrome.

As has been said, cancer is **very rare** in Noonan syndrome but one specific form of leukaemia has been reported:

- **Juvenile myelomonocytic leukaemia** is a very rare childhood blood cancer that occurs when immature blood cells (called blasts) make too many myelocytes and monocytes (two types of white blood cells) and cause a form of leukaemia. Fortunately there is now good treatment for this condition in specialised centres.

There are long term follow up studies being carried out to investigate whether people with Noonan syndrome have a higher incidence of other cancers in later life, but at present there is no evidence for this.

REFERENCES/FURTHER READING

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