Methods for the diagnosis of neuroblastoma malignancies

CSIC has developed a method for discriminating LR-NBs and HR-NBs that should facilitate the design of a personalized therapy for a patient with neuroblastoma, as well as an activator of a gene belonging to the SA-c1 module for use in the treatment and/or prevention of neuroblastoma malignancies.

Industrial partners from the pharmaceutical or diagnostic industry are being sought to collaborate through a patent licence agreement.

Transcriptional landscapes of LR-NBs and HR-NBs can be discriminated based on the SA-c1 signature

Neuroblastoma is the most frequent solid extracranial tumor in childhood, accounting for 10-15% of cancer-related child deaths. NBs are nearly all sporadic (98%) and present a high degree of heterogeneity.

They can be classified as low-to-intermediate risk tumours (LR-NBs), which correspond mostly to tumours that present a good prognosis and can even regress spontaneously, or as high-risk tumors (HR-NBs), which show poor prognosis due to metastasis, strong resistance to current therapy and a high probability of relapse. Identifying the molecular signatures that discriminate LR-NBs from HR-NBs should facilitate the design of efficient therapies against aggressive NB tumours.

Transcriptional signatures associated to the formation of both LR- and HR-NBs have been identified, which might have use as a method for the diagnosis of NB malignancies. In addition, genes comprising the SA-c1 module are proposed as possible therapeutic targets for the treatment of NB malignancies, particularly HR-NBs.

Main innovations and advantages

- It has been demonstrated that the transcriptional landscapes of LR-NBs and HR-NBs can be discriminated based on a module of genes expressed by the core sympato-adrenal lineage (the SA-c1 signature), and that these genes are strikingly predictive of NB malignancy, patient prognosis and disease progression.
- A set of genes has been identified, which are expressed at significantly lower levels in HR-NBs than in LR-NBs, so that these genes might impede the formation of metastases specifically observed in HR-NBs.
- The activators are particularly helpful for NB patients showing at least one metastasis, and for those patients affected by NB tumours displaying a strong resistance to conventional therapy.

Heatmap representation of the differentially expressed genes (DEGs) comparing HR vs LR

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