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Title:

NASAL POTENTIAL DIFFERENCE OUTCOMES SUPPORT DIAGNOSTIC DECISIONS IN CYSTIC FIBROSIS

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What was your research question?

This study was designed to identify whether or not the Nasal Potential Difference (NPD) measurements are accurate in distinguishing a patient with cystic fibrosis (CF) versus a non-CF subject, and the most appropriate interpretation of this test performed on the mucous membrane of the nose for supporting diagnosis if the diagnosis is uncertain.

Why is this important?

Mutations of Cystic Fibrosis Transmembrane conductance Regulator (CFTR) gene causes CF. In atypical forms of CF as CFTR related disorders affecting only a single organ, the most common diagnostic tests, sweat chloride and genotype are often inconclusive; however complementary diagnostic tools can support uncertain diagnoses, such as NPD. The most widely used in vivo functional and intestinal current measurements is utilising rectal biopsies. In using these methods, it is essential to refer to the latest diagnostic Standard Operating Procedures (SOPs) available from the European Cystic Fibrosis Society (ECFS). Moreover, it is favourable to apply a common interpretation applicable to the results of the tests.

What did you do?

We applied NPD ECFS-SOP to 65 people with CF, 34 people without CF and 16 healthy carriers considered as reference group and to 81 subjects with uncertain diagnosis. Both nostrils were explored for the site of the most negative voltage using a catheter, delivering molecules affecting ion transport through the cell membrane. Activation or inhibition of ion channels as CFTR on the nasal mucosa was detected as variation of potential difference. The

specific responses to each molecule were considered in formulae previously proposed for calculating “Sermet score” and “Wilschanski index”. These results were all taken into account when presenting the outcome.

What did you find?

We found that both formulae are more appropriate than single responses for interpreting NPD results, when distinguishing patients with CF from non CF counterparts. Values of both "Sermet score” and “Wilschanski index" aligned with the final diagnosis of controversial cases, and the latter performed slightly better than Sermet score.

What does this mean and reasons for caution?

We concluded that it is very important to apply both formulae for appropriate and common interpretation of NPD results. Studies on this field from other centres would be very useful for confirming the validation of these formulas and for sharing data and discussing controversial cases.

What’s next?

This study is paving the way to achieve a common agreement on interpretation of NPD measurement. Important efforts are currently dedicated to this aim by several groups, focusing on standardization of quantification of CFTR function in human airways. This data could support clinicians in receiving approval for running this test in difficult diagnostic cases.

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