

Gene defects linked to eczema, wheeze and nasal disease among babies

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A link has been discovered between a common gene defect and eczema, nasal blockage and wheeze among babies as young as six months, according to a new study at Brighton and Sussex Medical



School (BSMS). The research raises new questions about how soon in life these defects could start affecting babies, resulting in serious health problems, and suggests treatment targeted towards children carrying these genetic defects started soon after birth could improve their lives.

The protein filaggrin is present in the skin and nasal cavity, helping to maintain the skin barrier, and previous studies have shown defects in the gene synthesising filaggrin are strongly linked to the risk of developing eczema and how serious the eczema and asthma turned out to be over childhood.

Professor Somnath Mukhopadhyay, Chair in Paediatrics at BSMS, said: "In the first study of this kind, we recruited mothers in the antenatal clinic and followed up the <u>babies</u> through infancy, in order to define the link between these gene defects, the resultant skin problem, and eczema, wheeze and nasal blockage, in early infancy. Our striking finding establishing this link could mean that some babies with these gene defects could be getting primed from birth or soon after, for a life of suffering from allergy-related disease."

The GO-CHILD study recruited 2,312 <u>pregnant women</u> in England and Scotland, who gave a cord blood sample at birth or saliva in infancy for genotyping of the babies. Babies were followed up for symptoms related to allergic diseases such as dry skin, eczema, wheeze and nasal blockage, at the ages of 6, 12 and 24 months by postal questionnaires sent to the carers.

The gene defects made eczema, wheeze and nasal blockage worse at six months. While the defects were affecting eczema at one year, they were no longer worsening wheeze and nasal blockage at this age. At two years, they were principally worsening eczema and nasal blockage, but not affecting wheeze. "The problems affecting the child change over time, filaggrin gene variation representing one major



ensemble within the grand orchestra of allergic disease," said Professor Mukhopadhyay.

He added: "The use of simple emollients from birth targeted towards those who have these gene defects may help correct this problem, thus alleviating suffering in infancy and also through life. This new approach for targeting treatment according to genetic information is known as precision or personalised medicine and a future trial could represent the first application of this exciting and novel approach in little babies otherwise primed to develop chronic disease from an early age.

"This skin barrier defect leads to selective entry of allergens to increase the burden of disease. Whether such barrier defects could make us more vulnerable to agents causing infection, such as viruses, has not yet been addressed. We are in the midst of a pandemic where some people are affected but many are not. Could a defective outer barrier in skin and mouth be making some of us more vulnerable? If so, would identifying patients carrying these defects help us better protect those that are more vulnerable?"

More information: Kaninika Basu et al, Filaggrin gene defects are associated with eczema, wheeze, and nasal disease during infancy: Prospective study, *Journal of Allergy and Clinical Immunology* (2020). DOI: 10.1016/j.jaci.2020.02.036

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