Study shows an association of early onset eczema in infants <3 months with a gene mutation predisposing to increased water loss through the skin

A recent pilot study found that filaggrin loss-of-function (FLG) mutations are associated with eczema and skin barrier impairment at 3 months of age. It also showed that skin barrier impairment was present in FLG mutation carriers even without clinical eczema. This study was undertaken on the same associations but on a much larger population of three month old babies.

405 infants were examined for eczema. Disease severity was determined by SCORAD eczema severity score. Transepidermal water loss (TEWL) was measured on unaffected forearm skin. Venous blood samples were screened for the four commonest FLG mutations in the UK white population (R501X, 2282del4, R2447X, and S3247X). Median SCORAD and TEWL measurements in children with and without eczema and FLG mutations were compared.

Dr Glenis Scadding, President of the British Society for Allergy and Clinical Immunology (BSACI) said ‘It has been known for some time that mutations in the gene for filaggrin, a protein involved in skin barrier function, can lead to severe dry skin due to failure of the epidermal barrier to prevent water loss. This important study shows that infants who are carriers of this genetic mutation are predisposed to eczema due to increased water loss through the skin even before clinical signs of eczema appear. This has important implications for the future prevention and management of eczema in infants.

The results showed 22% (90/405) of children had clinical eczema. Median SCORAD was 10.7 (range 3.5-49.4). TEWL was higher in children with eczema compared to unaffected infants (median TEWL [g/m2*h] 16.84 (IQR 12.23, 23.84) vs 12.65 (10.41, 15.30), p<0.001. Higher TEWL was associated with more severe disease (r=0.50, p<0.001, median TEWL SCORAD <15 15.8 (12.0, 20.7) vs SCORAD >15 25.4 (15.0, 37.4), p<0.001). 13% (52/405) of children carried at least one FLG mutation. FLG mutation carriers were more likely to have eczema by three
months of age (OR=3.39 (1.84-6.22), p<0.001). FLG mutations were significantly associated with higher median TEWL (all children FLG 24.14 (17.02, 31.93) vs FLG 15.17 (11.95, 20.67) p<0.001), even without clinical eczema (FLG 15.10 (13.04, 19.91) vs FLG 12.51 (10.28, 14.90), p=0.01).

Already at three months, FLG mutations are associated with eczema phenotype, dry skin and transepidermal water loss. The observation that TEWL is elevated in unaffected FLG mutation carriers suggests that skin barrier impairment precedes clinical eczema.

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References
Flohr, C1; Logan, K1; Marrs, T1; Radulovic, S1; Barker, J2; McLean, I3; Lack, G1; Perkin, M1
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Notes to Editors
This study will be presented during the BSACI Annual Meeting which takes place from 11th -13th July at the East Midlands Conference Centre, Nottingham, UK.

About The British Society for Allergy and Clinical Immunology
The British Society for Allergy & Clinical Immunology (BSACI) is the national, professional and academic society which represents the specialty of allergy at all levels. Its aim is to improve the management of allergies and related diseases of the immune system in the United Kingdom, through education, training and research.

The BSACI website hosts the only comprehensive list of the NHS allergy clinics in the UK which BSACI actively encourages GP’s to use when referring patients to an allergy specialist for treatment. www.bsaci.org

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