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| Clinical features of children with cystic fibrosis screen positive indeterminate diagnosis [CFSPID] at Monash Children’s Hospital |
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| **Introduction/Aim:**  Newborn bloodspot screening [NBS] for cystic fibrosis [CF] usually allows early diagnosis of CF. However some infants with a positive NBS have equivocal results on subsequent gene mutation analysis and sweat testing. A diagnosis of Cystic Fibrosis Screen Positive, Indeterminate Diagnosis [CFSPID] is made if: 1.The sweat chloride is normal [<30 mmol/L] with two CF mutations [at least one of which has unclear clinical consequences] or 2.The sweat chloride is intermediate [30-59mmol/L] with one or no CF mutations.  Previous studies have shown that between 6% and 48% of these children are subsequently reclassified as CF [if the second mutation is found to be pathogenic] or converted to a CF diagnosis [a repeat sweat test become positive i.e. > 60 mmol/L].  This single centre case series examines a cohort of patients with CFSPID to review their phenotype and rates of reclassification or conversion over time.  **Methods:**  Children with CFSPID were identified retrospectively by review of medical records at the CF centre at Monash Children’s Hospital, Melbourne. NBS values, sweat chloride tests, CF gene mutations and clinical features of these patients were recorded.  **Results:**  Seven patients were identified with CFSPID, following an elevated IRT and an initial equivocal sweat test [Range 32-55 mmol/L]. Six patients had 1 copy of the F508 mutation and all had normal faecal elastase levels. Three patients subsequently converted to a diagnosis of CF following an elevated sweat chloride. The age at conversion varied widely from 11 months to 7 years of age.  **Conclusion:**  This case series highlights the importance of repeated sweat tests and follow up of CPSID cases as per recent guidelines1. More Australian data is needed to evaluate genotype, phenotype and rates of conversion to CF.  **Grant Support:** N/A  **Key words:** CFSPID, Cystic Fibrosis  Barben J, et al. Updated guidance on the management of children with cystic fibrosis transmembrane conductance regulator-related metabolic syndrome/cystic fibrosis screen positive, inconclusive diagnosis (CRMS/CFSPID). J Cyst Fibrosis. 2021 Sep;20(5):810-819. |