Conclusions This study’s main application lies in formulating interventions related to acknowledging misconceptions and stigmas related to Epilepsy and improving health-care systems by creating a team-based approach to patient care in meeting the required demands. These interventions may also be linked to devise applicable policies to impact reforming target behavior significantly.

235 GENOTYPE/PHENOTYPE CORRELATIONS IN 125 CHINESE PATIENTS WITH TUBEROUS SCLEROSIS: A 29 YEARS’ EXPERIENCE IN HONG KONG
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Background Tuberous Sclerosis Complex (TSC) is a multisystemic neurocutaneous disorder of autosomal dominant inheritance, with characteristic presentation of benign hamartomatous lesions across the brain, kidney, heart, eyes, skin and lungs.

Objectives To study the mutation spectrum and phenotypic characteristics of TSC patients in the Chinese population, as well as to delineate the underlying genotype-phenotype correlations and compare with previous studies.

Methods 120 patients suspected of TSC were referred to the Clinical Genetic Service (CGS), Department of Health, HKSAR for evaluation between 9/1991 and 8/2020. Blood samples from 105 patients presented with ≥1 major/ ≥2 minor diagnostic features according to the latest diagnostic criteria [1,2] are taken. Comprehensive genetic testing including sequencing and Multi-ligand probe-dependent Amplification (MLPA) was performed. Family screening was performed on molecularly confirmed cases. In total, 133 patients had a definite diagnosis, in which the 125 Chinese patients are included into the study. Statistical analyses (Pearson χ2 tests, Fisher exact test and ANOVA) are performed using SPSS version 26.0.

Results Pathogenic genetic alternations are identified in 72.0% patients (90/125), in which 26.4% (33/125) have TSC1 and 45.6% (57/125) have TSC2 mutations. 28 novel mutations are reported while familial cases account for 23.2% (29/125). Males have significantly more subependymal nodules (47/55 vs 34/50; p=0.033) than females, whereas de novo cases have more cortical tubers (69/82 vs 14/24; p=0.007) and renal angiomyolipoma (44/85 vs 3/21; p=0.002) than familial cases.

TSC2 cases have more frequent mental retardation (29/42 vs 4/25; p<0.001), cardiac rhabdomyoma (21/45 vs 3/25; p=0.003), renal angiomyolipoma (24/49 vs 5/27; p=0.009) and facial angiofibromas (37/50 vs 16/31; p=0.039) than TSC1, while mutation-negative cases show less subependymal nodules (14/23 vs 65/77; p=0.015) than mutation-positive cases. Similarly, TSC2 cases show higher occurrence of subependymal nodules (44/50 vs 14/23; p=0.012) than mutation-negative cases though mutation-negative patients have more frequent mental retardation (10/21 vs 4/25; p=0.020), autism spectrum disorder (4/18 vs 0/23; p=0.030) and renal angiofibromas (12/23 vs 5/27; p=0.012) than TSC1 cases. There are no significant phenotypic differences between patients with missense and protein-truncating mutations in TSC2, while TSC2 missense mutations are associated with more mental retardation (17/21 vs 4/23; p<0.001), cardiac rhabdomyoma (11/21 vs 2/24; p=0.001), renal angiomyolipoma (13/23 vs 4/24; p=0.004) and renal cysts (6/23 vs 1/24; p=0.048) than those with TSC1 protein-truncating mutations. Mutations in the coiled-coil region (TSC1) are significantly associated with nail abnormalities. All 14 antenatal-onset patients have cardiac rhabdomyoma. Meanwhile, they have less seizures (6/11 vs 55/64; p=0.027) than paediatric-onset cases but have higher frequencies of mental retardation (5/11 vs 0/11; p=0.035) than adult-onset patients. Generally, paediatric-onset patients have more neurological manifestations, while initial presentations of adult-onset TSC are more diverse. The predictive yield of antenatal cardiac rhabdomyoma on TSC is 63.6%.

Conclusions The overall phenotypic spectrum and genotype-phenotype correlations in our Chinese cohort are compatible with literature. 28 novel mutations have been reported in this study.

Abstract 234 Table 1

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KNOWLEDGE, ATTITUDES AND PRACTICES TOWARDS BREASTFEEDING AMONGST PAEDIATRIC HEALTHCARE PROFESSIONALS

Mercy Murinye Magwenzi. UK
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Background The World Health Organisation recommends exclusive breastfeeding of infants for six months when complementary feeds are introduced whilst continuing breastfeeding for up to two years of age and beyond. Ongoing predictable support to breastfeeding mothers is essential to improve breastfeeding rates and paediatric hospital doctors and nurses have an opportunity to provide this support when breastfed infants are admitted to their wards.

Objectives This study aimed to assess the breastfeeding knowledge, attitudes, and practices amongst paediatric healthcare professionals in a large teaching hospital in the UK.

Methods We conducted a qualitative study through a self-administered online questionnaire. Participants were doctors, nurses, nursery nurses and student nurses working in the paediatric department at the time of the study. There were 32 respondents, 14 of these were doctors.

Results

- Emergent themes were identified and analysed across all interviews. Five themes emerged: Breastfeeding knowledge and training, the influence of paediatric work experience, practitioner attitude towards breastfeeding, role in breastfeeding and practice in supporting breastfeeding.
- Almost all the participants had good levels of breastfeeding knowledge though only 50% of doctors had received formal breastfeeding training and were relying on personal experience whilst 70% of nurses had received training.
- All participants had a positive attitude towards breastfeeding with nurses mostly indicating comfort in the role of supporting mothers. A significant proportion of doctors indicated a lack of conviction in their ability to support breastfeeding mothers.
- Inconsistent advice would be offered to mothers about bottle feeding by different practitioners.

Conclusions 7 out of 14 doctors had no breastfeeding training and doctors were found to be less confident in the role of supporting breastfeeding mothers. There was good knowledge base amongst all the healthcare professionals however the actual practice of advice offered in supporting mothers was inconsistent amongst practitioners.

We hope that the results of this study will encourage formal breastfeeding education to be offered for all health care professionals working in paediatrics and the development of departmental breastfeeding policies to enable consistency whilst improving the care offered to patients and their families.