

1861 YOUNG PEOPLES' PERCEPTION OF SYSTEMIC DISEASE RISK IN PRIMARY CARE

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Objectives Systematic Patient and Public Involvement and Engagement (PPIE) aimed to discover the views of young people (YP) on informing patients and their families of the risk of systemic disease (systemic lupus erythematosus (SLE), inflammatory bowel disease (IBD) and Behcet's) given specific symptoms and demographics, in a primary care setting. Whilst this research directly informed a cohort study of primary care data assessing the risk of systemic disease in children and young people who presented to primary care with mouth ulcers, the findings are applicable to the wider primary care context.

Methods A multi-phase approach was adopted to address the aim. An initial meeting with eight members of GenR Liverpool (a group for young people interested in contributing to health care research), established important outcomes and language to be used in the project as a whole. Next, an online survey was distributed via social media posts and mailing lists facilitated by various charities involved with each systemic disease (SLE, IBD and Behcet's). A total of 218 responses provided further insight into the perception of proposed outcomes and language from both patients and parents. Finally, a meeting with six YP involved with YourRheum (a young person's advisory group for patients with rheumatic conditions) discussed the results of the primary care cohort study in terms of acceptability and dissemination of the findings.

Results In the first phase meeting, YP felt that it may be reassuring and empowering to know the systemic disease risk associated with their symptoms and demographics. However, they stressed that there must be a balance between 'realistic and scary' when communicating risk, and that knowledge is only helpful if there is action that can be taken.

In the second phase, 'red flag' information and advice on symptomatic treatment were deemed to be most beneficial at the early stages of diagnosis. YP and their parents felt that a risk calculation would: raise awareness of rare conditions; reduce time to diagnosis; allow preparation time; validate presenting symptoms; provide reassurance; and potentially allow earlier action to be taken. Concerns regarding risk calculation in primary care included: oversimplifying complex presentations; causing unnecessary worry; the risk of misdiagnosis; and the potential to demise patients and their families.

In the final phase meeting, results of the primary care mouth ulcer cohort study were presented and it was deemed that since the risk was 'low' (in this study) it would be unhelpful to be informed of the exact risk of systemic disease given the presence of mouth ulcers. However, some patients/parents may be reassured. Nevertheless, since the COVID-19 pandemic, YP with experience of long-term health conditions were keen to highlight the mental health impact of being labelled 'at risk'. YP preferred age-appropriate, positive language, ideally communicated 'face-to-face' by consultants, when receiving information about risk.

Conclusions Overall this PPIE research provides insight into the thoughts, feelings and opinions of YP around the risk assessment of systemic disease in primary care. These views must be considered in any aspect of practice that involves early stage diagnosis or uncertainty.

1865 THE IDENTIFICATION OF DE-NOVO PATHOGENIC GENETIC MUTATIONS IN ADOLESCENT PATIENTS WITH ADHD

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Objectives This study analyses a sample of adolescents in Qatar with attention deficit disorder, to find associated pathogenic genetic mutations using whole genome sequencing.

Methods A sample of 14 families of adolescents with ADHD was investigated in this study. The total number of participants was 84 people. That included affected patients with ADHD and their parents. Some of the patients' healthy siblings were included for comparison. The patients attended the adolescent medicine clinic in Sidra hospital, Doha. The age group of the participant patients was between 12 and 18 years, the majority of them were males, and apart from ADHD, they were healthy with no underlying neurological diseases. They were diagnosed with ADHD by consultants in adolescents' medicine based on their clinical presentation and DSM-5 criteria,¹ supported by their scores in the Vanderbilt questionnaire, which was filled by their parents and teachers. Whole blood samples from the participants were sent for genetic study using whole exome sequencing (WES). Results of these tests were carefully studied for genetic mutations, and compared with their parents' results to check whether these mutations were inherited from one or both parents or occurred sporadically.

Results This study has identified pathogenic de-novo variants in 5 genes (*HERC2*, *FARP2*, *GRIA2*, *SMURF1*, and *TUBB3*), in 5 probands out of 14 families (table 1). These genes are expressed in the brain, and they have clear associations with some neurological conditions. The *HERC2* gene mutation is associated with autism spectrum disorder in childhood, the *FARP2* is associated with Brachydactyly-Mental Retardation syndrome, the *GRIA2* is associated with Psychiatric and Neurodevelopmental Disorders, the *SMURF1* is associated with Speech-Language disorder and the *TUBB3* is associated with cortical dysplasia with other complex brain malformations.

Abstract 1865 Table 1

Chromosome\ Gene	Variation Effect	Brain regional specificity	Clinical Variant Pathogenic status
15\HERC2	Splice	Cerebellum	Pathogenic
2\FARP2	Splice	Cerebellum	Pathogenic
4\GRIA2	Splice	Cerebral cortex and basal ganglia	Pathogenic
7\SMURF1	Splice	Cerebral cortex, Pons, Medulla	Pathogenic
16\TUBB3	Splice	Hypothalamus, Pons, Medulla	Pathogenic

Conclusions Attention-deficit hyperactivity disorder (ADHD) is a common childhood-onset neurodevelopmental disorder characterized by inattention, impulsivity, and hyperactivity. ADHD exhibits substantial heritability, with rare monogenic variants contributing to its pathogenesis. ADHD prevalence in Qatar is considered significantly high, with almost 10% of the Qatari children suffering from ADHD.² This could be linked to the fact that the rate of consanguinity amongst the Qatari population is particularly high reaching up to 54%.³

This study has found five de novo genetic mutations in five families of patients with ADHD, supporting the evidence of the genetic basis of ADHD. Identifying these mutations will contribute to the future of precision medicine, which will allow doctors around the world to tailor their management based on their patient's unique genetic characteristics, for accurate diagnosis and optimum care.

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CLINICAL OUTCOMES FOR ADOLESCENTS LIVING WITH HEPATITIS B

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Objectives 1) To audit clinical outcomes for adolescents living with chronic hepatitis B (CHB)

2) To audit clinical outcomes following transition to adult services

Methods Retrospective case note analysis extracting a dataset of all patients seen in the paediatric CHB clinic between 2010 and 2022. Data collated: e-antigen (eAg) status; e-seroconversion; alanine aminotransaminase (ALT); coinfection with hepatitis D virus (HDV), hepatitis C virus (HCV) or HIV; liver inflammation and fibrosis by transient elastography and biopsy; and treatment status for hepatitis B (HBV).

Results 58 children, 36 (62%) male, presented to paediatric CHB care. The median age at presentation was 13 years (IQR 6, 15). Ethnicity: Asian 25 (43%), black African 15 (26%), White 8 (14%), Other 3 (5%), Unknown 5 (9%). Viral coinfection: HDV (1), HIV (1) and HCV (0). 21 (36%) transitioned to adult services at a median age 18 years (IQR 18, 19), median length of time in adult care 4.6 years (IQR 3.1, 5.5), with no loss to follow up.

At latest assessment; median age 18 years (IQR 12, 22); median ALT 38IU/L (IQR 24, 49) in paediatric care (n=37) and 44IU/L (IQR 29, 65) post-transition. 5 have ALT > twice upper limit of normal: 3 paediatrics; 2 adult care. 33/58 (57%) are eAg negative, 11 e-seroconverting during follow-up: in paediatrics (10) and post-transition (1). Median HBV DNA by HBeAg: positive 87,000,000IU/mL (IQR 20,996, 641,000,000); negative 235IU/mL (IQR 33, 1537). Latest transient elastography mean CAP 198dB/m (SD ±57), mean E score 5.1kPa (SD ±1.7) with 5 having evidence of mild or severe fibrosis (F2 E score 7.4–11.1)

19 (33%) ever received HBV therapy, median age 14 years (IQR 8.5, 15); 1 treated post-transition aged 22. The pre-treatment median ALT 47IU/L (IQR 28, 64); median HBV

DNA 401,242IU/mL (IQR 635, 226,500,000). 15 received pegylated interferon alpha (PEG-IFN α); clinical trial (3), adult care (1). 3 stopped due to toxicity, including 1 post-transition. 4/15 (27%) e-seroconverted.

4/19 received tenofovir disoproxil fumarate: median age 14 years (IQR 14, 15); median HBV DNA 85,003,498IU/mL (IQR 6295, 579,700,000), with 2 achieving sustained viral suppression. 2/4 adolescents were eAg negative from treatment initiation.

11/58 underwent liver biopsy all pre-2018 in paediatric care. The modified Hepatic Activity Index (HAI) necroinflammatory scores median was 3/18 (range 1–8). The HAI fibrosis stage median 1/6 (range 1–3).

49/58 underwent transient elastography in paediatric services; mean CAP score 197dB/m (SD ±44), mean E score 5.4kPa (SD ±1.3). Three children had E scores >7.4kPa; all received treatment. Post-transition, 9/21 underwent transient elastography; mean CAP score 246dB/m (SD ±49). The mean E score was 5.9kPa (SD ±2.3) with two having E scores >7.4kPa; none has been treated.

Conclusions In this cohort of adolescents living with HBV rates of cirrhosis were reassuringly low. More than half had undergone e-seroconversion, all but one prior to transition to adult care. One third e-seroconverted during follow up, the majority spontaneously. PEG-IFN α did not induce e-seroconversion in most cases when used.

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THE CHALLENGES OF THE COVID-19 PANDEMIC ON YOUNG PEOPLE WITH EATING DISORDERS: OUR EXPERIENCE IN QATAR

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Objectives This study examined the impact of the COVID-19 pandemic on adolescents with eating disorders, mainly anorexia nervosa, and Bulimia nervosa, using data collection pre-pandemic and during the pandemic in the outpatient clinics in Sidra hospital, Qatar.

Methods Medical records of the patients with eating disorders were reviewed for the period between August 2017 and April 2022. Diagnosis of Anorexia Nervosa and Bulimia Nervosa was done using the DSM-V criteria. For the purpose of this study, August 2017 to March 2020 is considered 'pre-pandemic' and April 2020 to April 2022 is 'post-pandemic'.

The clinical assessment in the pre-pandemic period was carried out face to face in the clinic, while a hybrid model of clinical care that uses telephone consultations and a limited number of patients' physical appointments was adapted during the pandemic period. The number of clinical appointments increased gradually as the number of COVID-19 cases decreased in the country.

The study compared the numbers of diagnosed patients with eating disorders between the two mentioned periods and investigated their specific characteristics (including age, gender, and specific type of eating disorder) and associated comorbidities, like depression and anxiety.

Results In the pre-pandemic period, 58 adolescents aged between 8–18 years old were assessed and diagnosed with an eating disorder. Out of the 58 diagnosed with an eating disorder, 16 patients were diagnosed with co-morbid depression