RECOGNITION OF THE FOETAL AND PERINATAL FEATURES OF THE PRADER-WILLI SYNDROME IS REQUIRED TO AVOID DELAY IN DIAGNOSIS

[1]Filiz Mine Cizmecioglu, [2]Jeremy Huw Jones, [2]Wendy Forsyth Paterson, [3]Sakina Kherra, [4]Mariam Kourime, [2]M Guftar Shaikh, [5]Malcolm Donaldson.

[1] Kocaeli University, Paediatric Endocrinology & Diabetes Department, Turkey, [2]Royal Hospital for Children, Glasgow, United Kingdom, [3]Department of Pediatrics, CHU Parnet, Algiers, Algeria, [4]University Hospital Abderrahim Harouchi, Casablanca, Morocco, [5]University of Glasgow School of Medicine, Section of Child Health, Glasgow, United Kingdom

Introduction: Prompt diagnosis in Prader-Willi syndrome (PWS) is important for counselling the family and thus pre-empting the hyperphagic phase of the condition.

Objectives: To determine the key diagnostic features of PWS during the perinatal period and hence recommend strategies to ensure early diagnosis.

Study design: Retrospective case note review with prospective questionnaire survey of birth details for the affected child and healthy siblings in which mothers scored foetal movements on a scale of 1 (low) to 5 (high). Statistical analysis was performed with Minitab V.13

Results: Between 1991 and 2015 inclusive 90 subjects (54M:36F) with PWS were seen in the Glasgow multidisciplinary PWS clinic. Cause was paternal deletion (56), maternal disomy (26), imprinting centre mutation (2), translocation/deletion (1), mutation-negative (1), tested elsewhere (4)[*Figure 1*]. Mean maternal/paternal ages for disomic patients were 34.6/34.6 yr, significantly older than for deletion at 26.4/29.6 yr (*p<0.001 &* 0.004). Comparison of foetal movement score, gestational age, birthweight (both by gender and for the whole group) and prevalence of prematurity for disomic and deletion inheritance showed no significant difference.

PWS pregnancies featured polyhydramnios in 10/34 (29%), breech presentation in 15/53 (28%), and caesarean section delivery in 38/86 (44%). Median (range) birthweight and gestation for patients compared with siblings were 2.76 (1.18-3.99) compared to 3.3 (3.1-4.9) kg; and 39 (30-43) compared to 40 (33-42) weeks, with prematurity (<37 weeks gestation) in 21 (23.6%) and low birthweight (<2500g) in 28 (32%) of PWS patients.

Median (range) foetal movement scores were 1 (1-4) [n=80] for PWS compared to 3 (1-5) [n=94] for their siblings (p<0.001)[*Figure 2*]. Median (range) duration of nasogastric feeding and hospital stay was 30.5 days (2d - 1.3 yrs) and 27 days (0d - 2 yrs). Median (range) time to clinical/molecular genetic diagnosis (available ≥1991) was 2.5 m (1d - 46yrs) / 10 m (4d - 46.5 yrs). Stratifying by year of birth <1980, 1980-89, 1990-99, 2000-09, and >2010 showed significant improvement in median time (days) to clinical/molecular diagnosis: 1862/8395, 97/3577, 74/73, 19/60 and 7/14 days (p<0.001). However from 2000-2015 inclusive clinical diagnosis was > 28 days in 11 and > 1 yr in 5 patients.

Figure 1: Inheritance mode in 90 patients with PWS

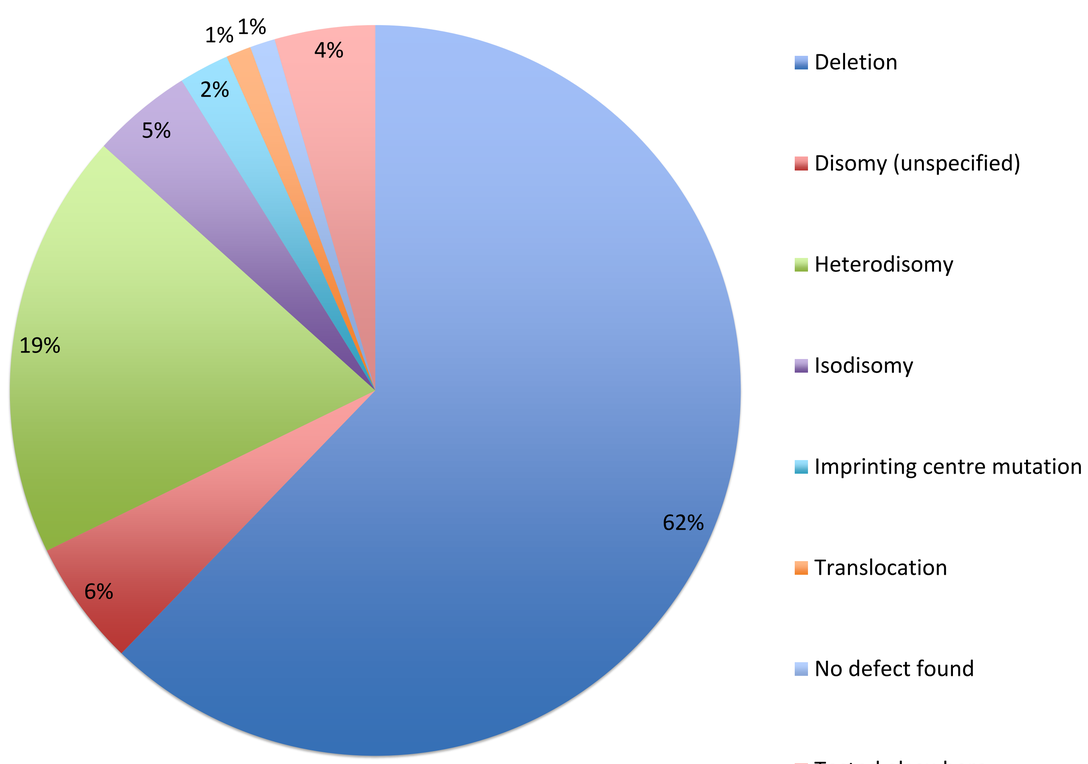
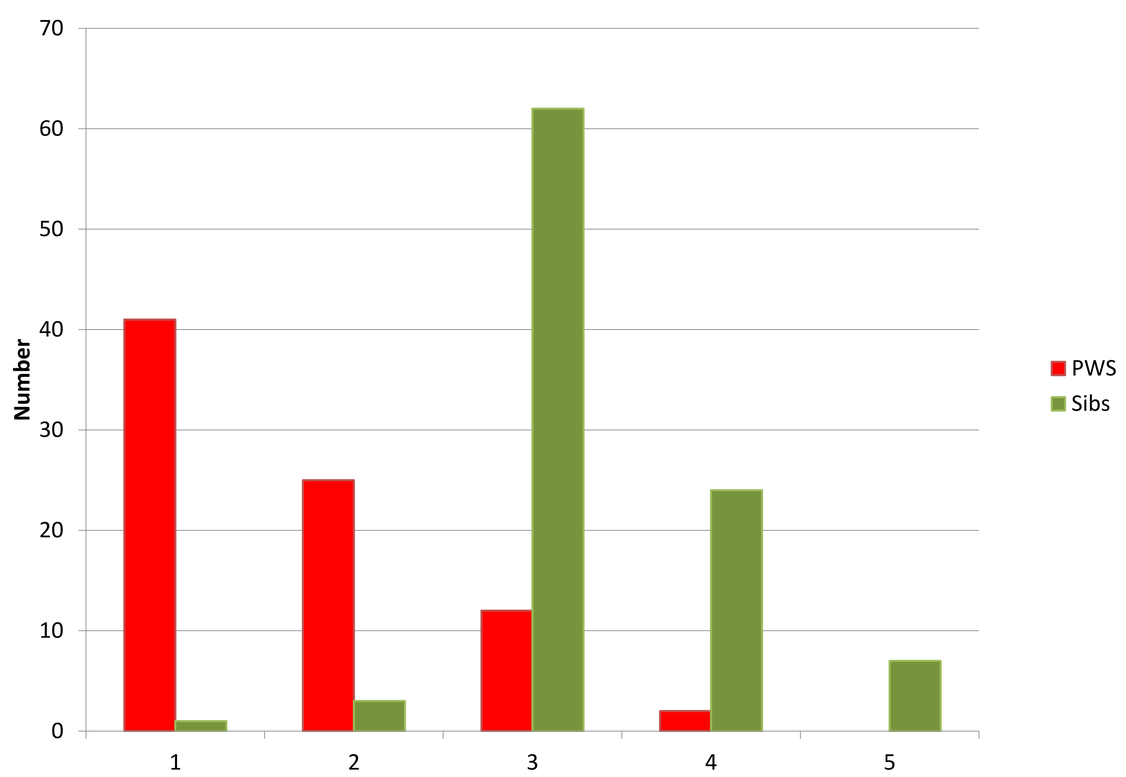


Figure 2: comparison of PWS and sibling foetal movement scores



Conclusions: Despite the overlap in features with prematurity, diagnosis of PWS can and should be made within days of birth if the key features which are mostly hypotonia-related but include cryptorchidism in males, are recognised and actively sought. Neonatal paediatricians need to be fully aware of the perinatal features of PWS, including a history of reduced foetal movements, so that unnecessary investigations in light of the severe hypotonia (such as brain MRI brain and muscle biopsy) can be avoided. Early diagnosis will also facilitate prompt initiation of familial counselling and education and therapy for the affected infant.

Conflict of interest: the authors have nothing to declare.

55ESPE 559--P1

Perinatal Endocrinology

Filiz Mine

DOI: 10.3252/pso.eu.55ESPE.2016

Poster presented at: ESPE 2016 PARIS

PosterSessionOnline.com