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CHALLENGING BEHAVIOURAL PROBLEMS IN CHILDREN WITH GENETIC AND RARE CONDITIONS: ROLE OF THE CANADIAN PAEDIATRIC SURVEILLANCE PROGRAM

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Background: With increased awareness, earlier diagnosis and management, researchers are unravelling medical, developmental and behavioural issues in patients with CHARGE association/syndrome, Prader-Willi syndrome (PWS), Smith-Lemli-Opitz syndrome (SLOS) and Early-onset eating disorders.
Aim: To collect descriptive information on natural history of these diseases and to identify underlying co-morbid diagnoses that can be more easily managed and some even prevented, with early intervention and careful long-term follow-up.
Methods: Using a monthly report form, over 2300 paediatricians and paediatric subspecialists actively report 10-12 different rare diseases according to preset definitions and protocols. Principal investigators analyze case-specific clinical data provided on follow-up questionnaires.
Results: CHARGE association/syndrome patients experience delayed puberty, obsessive-compulsiveness, socializing difficulties and sleeping problems. While 50% demonstrate self-abusive behaviour and aggressive outbursts, over 70% can be improved with behaviour modification drugs. Children with PWS exhibit such classic behavioural problems as skin picking, stubbornness, temper tantrums, anxiety, hoarding, food obsession and overeating. Early diagnosis enables the family to develop behaviour modification skills and start an effective Red-Yellow-Green Diet System. The behavioural phenotype of SLOS patients includes autistic features, sensorial hyperactivity, sleep disturbance, aggressivity and automutilation; however, treatment with dietary cholesterol supplementation alleviates these problems. Many behavioural problems detected in Early-onset eating disorder patients could engender new diagnostic classification criteria.
Conclusions: CPSP studies have provided insight into the 'basket of behaviours' associated with these rare conditions. Consequently, behavioural problems can be prevented and/or managed by the family and the paediatrician to improve the quality of life for these children.