Cancer Genetic Risk Assessment in Pediatric Oncology: Results and challenges of a Brazilian Experience

Authors:
Fernanda Teresa de Lima, MD,PhD (IOP-GRAACC-UNIFESP, São Paulo, Brazil); Camila Maida de Pontes, NP (IOP-GRAACC-UNIFESP, São Paulo, Brazil); Carla Renata Pacheco Donato Macedo, MD (IOP-GRAACC-UNIFESP, São Paulo, Brazil); Nasjla Saba da Silva, MD (IOP-GRAACC-UNIFESP, São Paulo, Brazil); Andrea Maria Cappellano, PhD (IOP-GRAACC-UNIFESP, São Paulo, Brazil); Eliana Maria Monteiro Caran, MD, PhD (IOP-GRAACC-UNIFESP, São Paulo, Brazil).

Backgroung: Childhood cancer predisposition is variable and the access of affected children to Genetic Counseling Services is limited, even in developed countries. **Hypothesis:** To describe patients seen for genetic cancer risk assessment and genetic counseling in a public academic pediatric oncology hospital in Brazil from 2013 to 2015 and challenges faced. **Methods**: Chart review of patients evaluated from January 2012 to December 2015 at the Oncogenetic Clinic of IOP-GRAACC-UNIFESP (Pediatric Oncology Institute – Support Group for Children and Adolescents with Cancer – Federal University of São Paulo, São Paulo, Brazil). **Results:** A total of 290 patients were seen, from 268 families, most males (150/51,7%) and over 5 years of age (194/66,9%). Main diagnoses were neurofibromatosis type 1 (84/29%), retinoblastoma (56/19,3%) and tuberous sclerosis (30/10,3%). The exact diagnosis was not concluded in 58 (20%) patients, classified as congenital malformation syndromes associated to cancer (31/10,7%) or cancer predisposition syndromes (27/9,3%). For patients with tumors and clinical suspicion of known malformation syndromes, follow-up guidelines were adopted (eg. 13 patients with medulloblastoma and suspicion of Gorlin syndrome; 19 patients with sarcoma or adrenalcortex carcinoma or plexus choroid carcinoma and suspicion of Li-Fraumeni syndrome). Most patients were seen more than once (160/55,1%). Among the challenges faced were a great number of patients waiting for an evaluation and remaining to be seen; exiguous human resources to perform genetic evaluations; lack of patients' and family members' knowledge about the objectives of evaluation and about their cancer family history; lack of availability of molecular testing in a public setting and impossibility of follow-up of adult family members at the same institution due to the fact that IOP-GRAACC-UNIFESP is a pediatric only institution. **Conclusions:** Cancer Genetic Risk Assessment in a Pediatric Oncology Hospital facilitated the identification of genetic syndromes in this group of patients, with improvements in management and adoption of preventive and surveillance procedures. There are a number of challenges to be faced, especially in a public hospital of a low-income country.