Autism Spectrum Disorder in Infancy and Early Childhood: The Model of the Centro de Estudos do Bebé e da Criança for Diagnosis and Therapeutic Intervention

A Perturbação do Espetro do Autismo na Primeira Infância: O Modelo do Centro de Estudos do Bebé e da Criança de Avaliação Diagnóstica e Intervenção Terapêutica

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ABSTRACT

Introduction: The Centro de Estudos do Bebé e da Criança in Hospital Dona Estefânia has organized a multidisciplinary model for children under three with suspected autism spectrum disorder, thus implementing the recent guidelines established by the Directorate General for Health. The aim of this study is to describe this model and case series.

Material and Methods: A retrospective descriptive study of observed children with suspected ASD. They were observed according to the model of the Centro de Estudos do Bebé e da Criança and DC:0-5™ classification, between January 2018 and September 2019.

Results: The study included 178 children. The average age at the initial assessment was 27 months. From the total sample, 116 children concluded the diagnostic sessions (axis I): Autism Spectrum Disorder/Early Atypical (36%), Developmental Language Disorder (18%), Other (19%). Factors of axes II, III, IV and V of DC:0-5™ were determinant for clinical diagnosis in 26%.

Discussion: Of 116 children, 36% were diagnosed with Autism Spectrum Disorder. This highlights the diagnostic challenge posed by neurodevelopmental disorders in early infancy. The sample shows that the characteristics of the relationship with the caregiver (axis II), presence of physical conditions (axis III), psycho-social stressors (axis IV) and developmental trajectory (axis V) have a significant clinical impact. In the future, the initial assessment should take place well before the age of 27 months because of the impact on prognosis.

Conclusion: This model is a pioneering approach in Portugal. It promotes a common approach of Child and Adolescent Psychiatry and Neuropediatrics/Developmental Pediatrics in early infancy. Moreover, it increases the diagnostic acuity of Autism Spectrum Disorders and early therapeutic intervention.

Keywords: Autism Spectrum Disorder/diagnosis; Autism Spectrum Disorder/therapy; Child; Infant; Portugal

RESUMO

Introdução: O Centro de Estudos do Bebé e da Criança do Hospital Dona Estefânia desenvolveu um modelo multidisciplinar de atuação na suspeita de perturbação do espetro do autismo na primeira infância, aplicando a recente norma da Direção Geral da Saúde. Pretende-se descrever a sua apresentação e casuística.

Material e Métodos: Estudo retrospetivo descritivo da série de crianças (< 3 anos), observadas por suspeita de perturbação do espetro do autismo, entre janeiro de 2018 e setembro de 2019, segundo este modelo e a classificação DC:0-5™.

Resultados: Foram observadas 178 crianças. A idade média da primeira consulta foi de 27 meses. Do total de crianças observadas, 116 concluíram as sessões diagnósticas (diagnóstico eixo I): perturbação do espetro do autismo/ perturbação do espetro do autismo atípica precoce (36%), perturbação do desenvolvimento da linguagem (18%), outros (19%). Em 26% dos casos, o quadro foi atribuído a fatores classificados em outros eixos.

Discussão: O diagnóstico de perturbação do espetro do autismo foi colocado em 36%, demonstrando o desafio diagnóstico das perturbações do neurodesenvolvimento na primeira infância. A casuística demonstra ainda que as características da relação com o cuidador (eixo II), a presença de condições físicas (eixo III), fatores de *stress* psicossociais (eixo IV) e a trajetória de desenvolvimento (eixo V) têm um impacto clínico significativo. É desejável a antecipação da idade de sinalização pelo impacto no prognóstico.

Conclusão: Este modelo é pioneiro em Portugal ao propor uma atuação conjunta de duas especialidades na primeira infância: pedopsiquiatria e neuropediatria/pediatria desenvolvimento. Este modelo de atuação melhora a acuidade diagnóstica e permite a intervenção terapêutica precoce.

Palavras-chave: Criança; Lactente; Perturbação do Espetro do Autismo/diagnóstico; Perturbação do Espetro do Autismo/tratamento; Portugal

INTRODUCTION

Autism spectrum disorder (ASD) is in fact a heterogeneous group of neurodevelopmental disorders globally with impaired social interaction and communication (verbal and nonverbal) in addition to the presence of repetitive behaviour patterns and/or restricted interests. With early onset in childhood, ASD can lead to significant limitations in child's

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emotional and social functioning, leading to persistent impairments.[1-3]

An early therapeutic intervention has a relevant influence on the outcomes of ASD.[4-10] It is well known today that the adoption of a relationally-based therapeutic intervention taking into account advances in neuroscience and the recognition of the importance of gene-environment interactions for neuronal organisation, allows for a positive progression, showing that the course of this disorder is not always chronic and associated with poor outcomes.[4,6-14]

In early childhood, ASD diagnosis is a difficult clinical challenge, not only due to developmental aspects but also due to symptom instability at this age.[3,4,9] Understanding and systematising the specificities of ASD and the whole group of neurodevelopmental and mental health disorders in early childhood requires a classification involving a multiaxial approach to these disorders, allowing for the diagnosis and differential diagnoses.[3,4,15,16]

This is the main reason for adopting the Diagnostic Classification of Mental Health and Developmental Disorders of Infancy and Early Childhood (0-5) DC:0-5™ Diagnostic Classification of Mental Health and Developmental Disorders of Infancy and Early Childhood (Portuguese translation) rather than any other classification, including DSM-5 Diagnostic and Statistical Manual of Mental Disorders and ICD-10 International Statistical Classification of Diseases and Related Health Problems, which are commonly used in other age groups and inadequate to be used in early childhood.[17-19]

As regards diagnosis, ASD is included in DC:0-5™ diagnostic classification, in line with DSM-5 and ICD-10, even though a new entity, early atypical autism spectrum disorder (EAASD) is also included, allowing for the identification of early signs of ASD in children aged between nine and 36 months.[3,17]

At least two communication/socialisation difficulties and at least one criterion regarding repetitive restricted behaviour with some levels of functional impairment are required for an ASAP diagnosis. Therefore, the presence of a smaller number of clinical criteria at an earlier age is required, when compared to the proposed ASD diagnosis, allowing for the identification of difficulties that may be the target of intervention from very early in life.[17]

In addition to the diagnostic criteria for clinical disorders described in axis I of the DC:0-5™, the multi-axial approach of this classification also includes (i) issues related to the relational context, in axis II; (ii) physical health conditions and considerations, in axis III; (iii) psychosocial stress factors, in axis IV, and (iv) skills and developmental stages, in axis V. In addition to identifying disorders and vulnerabilities, it is also possible to find in each axis the strengths that may enhance an intervention. Therefore, the characterisation of each axis has a relevant clinical impact and should be specifically considered in diagnosis, treatment and understanding of child's development.

Unlike most disorders for which there is already a well-established specific follow-up and treatment approach strategy, clinical guidelines for ASD considering the different age groups are only recently emerging,[20-25] including the Portuguese Directorate-General for Health (DGS) clinical guideline (NOC) called "Diagnostic Approach and Intervention in Autism Spectrum Disorder in Paediatric and Adult Age", published in April 2019.[26] This guideline recommends the standardisation of the approach to ASD nationwide, clarifying issues related to early identification, diagnostic assessment and therapeutic intervention. As regards the early identification and diagnostic assessment, a referral to a hospital specialty consultation for children with suspected ASD is recommended, involving a multidisciplinary team in the diagnostic decision, therapeutic intervention and follow-up. A first consultation within 30 days is required for the referral of children aged 0-3.[26]

The team should have training and experience in ASD and should include a paediatrician with experience in neurodevelopment and/or a neuropaediatrician and a child and adolescent psychiatrist. The involvement of psychologists, nurses, social workers, speech therapists and occupational therapists or senior technicians in special education and rehabilitation is also required. An individualised approach to each patient should be used, based primarily on relational, non-pharmacological therapy.[26]

In order to address these different clinical dimensions, in compliance with the DGS NOC and complying with deadlines, the *Centro de Estudos do Bebé e da Criança* (Centre for Baby and Childhood Studies) (CEBC) at *Dona Estefânia Hospital – Centro Hospitalar Lisboa Central* (CHLC) has developed a multidisciplinary model for the diagnostic assessment and intervention aimed at children aged 0-3 with suspected autism spectrum disorder.

Launched in 2017, the CEBC brings together the early childhood unit of the childhood and adolescence psychiatry department and the development department at the *Dona Estefânia* Hospital and was mainly aimed at the differentiated care of children up to the age of 5, giving priority to children aged 0-3, following a direct contact by parents or by medical referral (children 0-3).

MATERIAL AND METHODS

This was a retrospective descriptive study of a group of patients diagnosed with suspected ASD in early childhood (0 - 3) approached according to the CEBC model as regards the diagnostic assessment and therapeutic intervention.

As this is the presentation and description of a diagnostic model, no specific informed consent was required. However, it was considered as included in the analysis of health data for the purpose of diagnosis and healthcare improvement. Data collection was based on the patients' clinical records. All patients aged 0-3 referred for suspected ASD between January 2018 and September 2019 were included.

Personal data were coded to ensure participant confidentiality and privacy. The study was approved by the Ethics Committee of the CHLC.

Three child psychiatrists, one neuropaediatrician, three

paediatricians, two clinical psychologists, two specialist nurses (mental health and psychiatry), two specialist nurses (child health and paediatrics), one occupational therapist, one speech therapist, one social worker and one special education teacher were included in the CEBC team.

The CEBC model of diagnostic assessment and therapeutic intervention in patients aged 0-3 with suspected ASD included (Fig. 1):

1. Initial assessment in a developmental and child psychiatry consultation by specialist doctors and specialist nurses (mental health and psychiatry and child health and paediatrics);
2. Follow-up through 10 weekly sessions (child psychiatry). These sessions aimed at clarifying the differential diagnosis and in establishing a relational-based therapeutic intervention, based on the D.I.R./Floortime® model (model based on development, individual differences and relationship) adopted as model of choice for the treatment of ASD[6,9,14,27] (axes I, II and IV were therefore approached);
3. The tenth diagnostic session included a joint consultation involving child psychiatrists and neuropaediatrician/developmental paediatricians; the final picture was established at this stage, considering aspects related to clinical progression, differential diagnosis, physical health conditions and considerations (axis III of the DC:0-5™ classification), need for clarification and additional investigation and developmental skills (axis V of the DC:0-5™ classification);

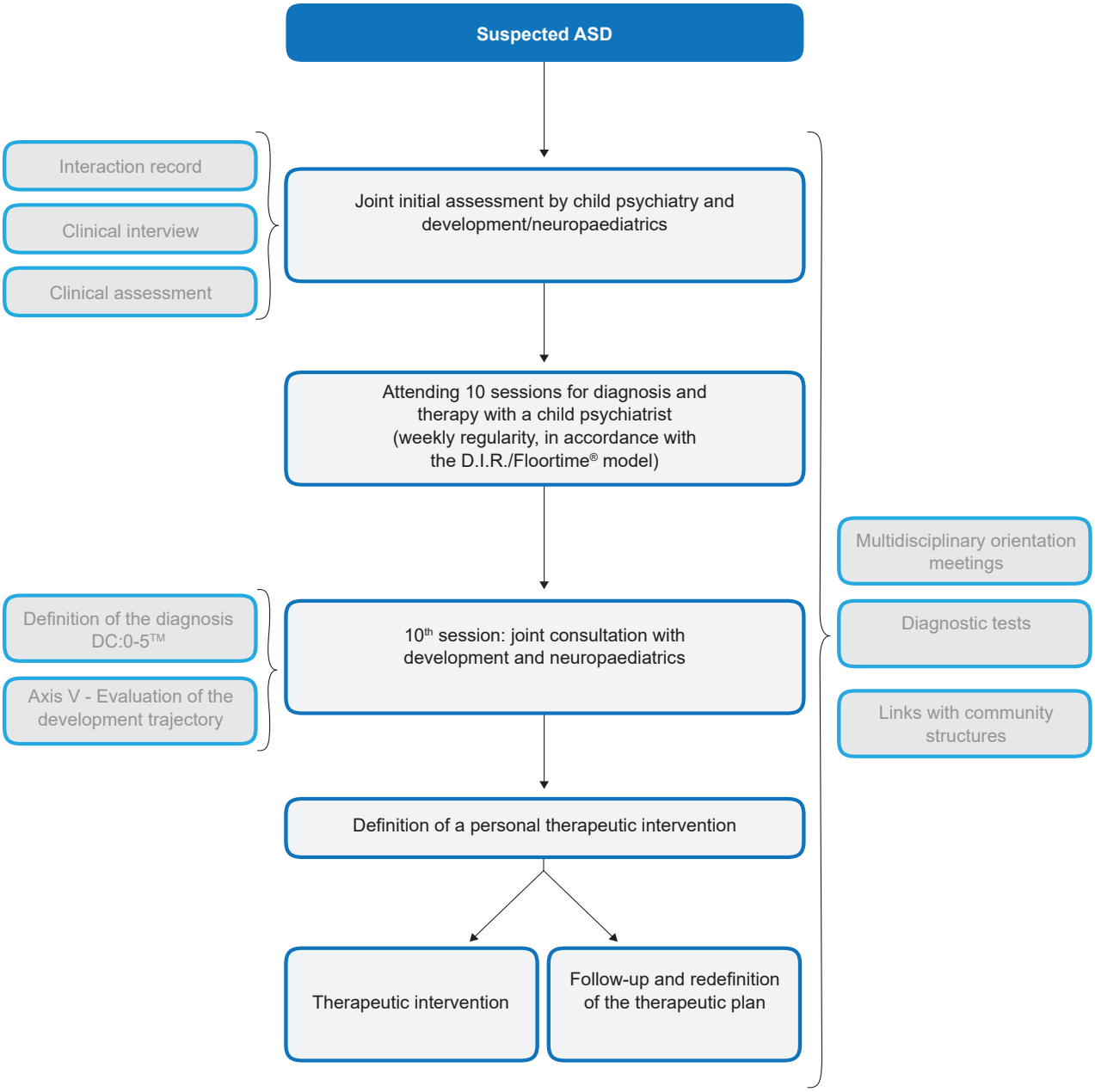

Figure 1 – CEBC model of diagnostic assessment and therapy in early childhood ASD

4. Therapy intervention carried out by occupational therapy, psychology or speech therapy teams in collaboration with community structures including nursery schools or kindergartens and local early intervention teams (*Sistema Nacional de Intervenção Precoce na Infância*);
5. Discussion of diagnosis and management strategy with the team in weekly clinical meetings (interaction observation meetings, supported by video records of the first consultation and clinical orientation meetings);
6. Identification of a reference professional (physician, nurse and/or psychologist, occupational therapist or speech therapist) who becomes the liaison element for the patient's follow-up after the diagnostic sessions.

RESULTS

A total of 178 patients aged 0-3 were referred throughout the study period with suspected ASD (135/76% male).

Patients were mainly referred by general practitioners (n = 69; 39%) or following the patient's family direct request (n = 48; 27%), while 34% of the patients were referred from other departments of the *Dona Estefânia Hospital* (n = 61), including medical paediatrics (13%), neuropaediatrics (7%), neurodevelopment risk outpatient clinic (4%), emergency (3%) and other departments (7%).

Delayed language acquisition and/or impaired social communication/interaction skills and/or the presence of a clear suspicion of ASD were the most frequent reasons for referral by physicians and families (total n = 160). The remaining reasons were related to behavioural disorders including tantrums or hyperkinesis, sleep and eating disorders considered within a wider context of suspected ASD from the first visit to the CEBC.

Patients were examined for the first time between the age of 9 and 36 months (average of 27 months), with a gradual increase of referred patients aged from 9 months onwards, reaching a maximum at 23 months (n = 12) and at 35/36 months (n = 26).

Regarding 25% of the patients (n = 44), both parents were immigrants originally from Nepal (n = 13), Brazil (n = 13), Angola (n = 7), Bangladesh (n = 4), Ukraine (n = 3), São Tomé and Príncipe (n = 2), India (n = 1) and Romania (n = 1).

A diagnostic discussion was carried out in 116 of the 178 patients, while the remaining patients are currently still attending diagnostic sessions.

The following diagnoses were proposed (n = 116): ASD, n = 30 and EAASD, n = 12 (total 36%), language development disorder, n = 21 (18%), global development delay, n = 12 (10%), others, n = 11 (9%) (example: post-traumatic stress disorder, mood disorder, sensory integration disorder). No diagnostic criteria for neurodevelopmental disorders were found in 30 out of these 116 patients (26%) (regarding axis I of the DC:0-5TM classification); therefore, the factors classified in axes II, IV and V of the DC:0-5TM (regarding the relational context, psychosocial stress and individual developmental trajectories) were considered as main determinants of the condition.

A management plan was established upon multidisciplinary diagnostic discussion (n = 116), including follow-up and therapeutic interventions at the early childhood department (40%) and at the development department (42%; 8% of these patients attended individual sessions for development promotion). During follow-up, 13% (n = 15) of the patients were transferred to other clinics within the patient's area of residence and all patients were referred to the local early intervention teams.

After the end of the diagnostic sessions, regular therapeutic support was provided to 40 patients at the speech therapy department (34%), occupational therapy – 17 patients (15%) and psychology (3 patients), attending a regular follow-up at the CEBC team. Five patients attended intensive therapeutic plans at the *Associação de Apoio à Unidade da Primeira Infância* (AAUPI). (Table 1)

Additional studies for aetiological investigation and/or genetic counselling to parents were carried out (25% of the patients; n = 45), including referral to genetics, metabolic study, neuroimaging study and electroencephalogram, whenever appropriate.

DISCUSSION

The differential diagnosis for children with ASD at this early age involves ruling out any other mental health and neurodevelopmental disorders, requiring technical expertise, the adoption of criteria and the consideration of axes that are not included in the DSM 5 classification and correspond to the core of the CEBC approach.

A total of 116 patients were included in the study upon having completed the diagnostic assessment for suspected ASD and 36% complied with the ASD/EAASD diagnostic criteria (Axis I DC:0-5^{TM}), reflecting the diagnostic challenge that it represents.

On the other hand, around 26% of the patients referred with suspected ASD were not diagnosed with neurodevelopmental disorders (Axis I DC:0-5^{TM}), showing the relevant impact of the elements considered in the other axes of the classification, namely the characteristics of the relationship with caregivers (axis II), the presence of physical conditions (axis III), psychosocial stress factors (axis IV) and the patient's own developmental trajectory, showing possible transient route deviations.

The assessment of the developmental trajectory (axis V DC:0-5^{TM}) during the initial diagnostic sessions and the CEBC follow-up model provides relevant data for the consideration and rationalisation of any additional diagnostic tests, that were obtained in about 25% of the patients, considering the presence of developmental regression, abnormalities in somatic examination or focal neurological signs, irritability or extreme apathy, impaired clinical progression or stagnation during sessions, comorbid conditions including epilepsy, the global involvement of the various developmental competencies or the parents' wish to attend genetic

Table 1 – Characteristics of the study population

Gender	**n = 178**
Female	43 (24%)
Male	135 (76%)
Referral	**n = 178**
General practitioner	69 (39%)
Direct family request	48 (27%)
Other departments	61 (34%)
Diagnosis (patients who have attended the 10 sessions)	**n = 116**
Autism spectrum disorder	30 (26%)
Early atypical autism spectrum disorder	12 (10%)
Language development disorder	21 (18%)
Global development delay	12 (10%)
Other diagnosis	11 (9%)
No Axis-I diagnosis	30 (26%)
Management plan (patients who have attended the 10 sessions)	**n = 116**
Early childhood department	46 (40%)
Development department	48 (42%)
Transferred to outpatient clinics within the patient's area of residence	15 (13%)
Intervention by local teams of early intervention	116 (100%)
Speech therapy	40 (34%)
Occupational therapy	17 (15%)
Psychology	3 (3%)
Multidisciplinary approach at the *Associação de Apoio à Unidade da Primeira Infância*	5 (4%)

counselling.[26,28,29]

It is worth mentioning that the CEBC follow-up model is guided by the reference physician and technician and, depending on the patient's clinical progression, it may be redesigned to include different CEBC departments. The therapeutic approach to ASD patients involved sessions based on the D.I.R/Floortime® model and progressive links with the community. The approach to the remaining patients was discussed on a case-by-case basis, depending on the patient's specificities, family needs and available resources within the community. In some cases, these involved the support of a nurse specialist, psychologist, occupational therapist and/or speech therapist. Globally, a similar distribution of the different follow-up approaches among the CEBC departments (early childhood and development departments) was carried out. The overlapping of performances and consultations is prevented by this flexible dynamic and favours family adherence by reducing family overload and the absenteeism of parents from work.

Therefore, we consider that this model offers a flexible response and that it has proved to be effective and efficient. This convenience sample showed our response to a very significant number of requests (n = 178) received within a short period (21 months); at the family request, direct access to very specialised care was made available, complying with the time limits (less than 30 days) recommended by the DGS NOC for the first consultation.

This model also aimed to privilege an early therapeutic intervention in children with suspected ASD and the expected impact on outcome improvement. The precocity of the intervention depends on the average age at the first consultation (27 months, in our group of patients). We believe that this could still be improved, as a referral peak at 23 months was found in this study, upon a progressive increase from the age of nine months onwards.

A trend towards an even earlier referral of the patients (earlier than 23 months of age) will hopefully exist as, on the one hand, major signs of ASD suspicion could be found within the first year of life and, on the other hand, a universal screening of this condition at the age of 18 months is recommended by the DGS National Program of Child and Youth Health (with the application of the M-CHAT / M-CHAT R/T questionnaire).[30-32] Therefore, awareness of a more timely referral should be reinforced among paediatricians, general practitioners, nurses in primary care teams and specialist nurses in paediatrics and child health, mental health and child and adolescent psychiatry.

It is also worth mentioning the presence of patients in this group born to parents of different foreign origins and cultures, corresponding to around 25% of the patients, showing the fact that parishes with the highest multicultural presence attend the *Dona Estefânia Hospital*.[33] Nine out of these patients were diagnosed with EAASD. The influence of immigration/multiculturality in the emergence of signs of

suspected ASD or its influence on the clinical progression of patients could not have been discussed with the available data obtained within this short period. However, we believe that immigration and multiculturalism could have increase social, family and economic vulnerability reflected in the characteristics of the relationships and in the presence of additional stress factors (axes II and IV DC:0-5™).

The approach to patients with ASD requires the access to direct and frequent intervention in occupational therapy, speech therapy and psychology, often requiring two or three of these simultaneously.[6,14,27] As regards this issue, due to staffing shortage, as mentioned in our results, there is a limitation in public care in speech therapy, occupational therapy or psychology, reflected in the low number of patients in our group who directly and regularly attended these approaches, according to our assessment.

CONCLUSION

The landmark CEBC model of diagnostic assessment and approach to patients with suspected ASD in early childhood (0 – 3 years of age) is to the best of our knowledge unique in Portugal as it promotes joint and often simultaneous approach by different teams including early childhood, childhood and adolescence psychiatry and development departments. The combined action between specialties and competences in compliance with the CEBC model, their follow-up and approach in weekly diagnostic sessions and regular team discussion (including the observation of recordings) contribute to the accuracy of the diagnosis of ASD in early childhood.

The use of DC:0-5™ (a multi-axial diagnostic classification of early childhood mental health and developmental disorders) in the diagnostic discussion allowed to consider the specificities of this age group, differential diagnosis and the identification of areas of approach to the strengths and vulnerabilities of each patient's functioning and relationships. In addition to diagnostic acuity, the CEBC model also gives direct access to very specialised healthcare, at the request of the family (i) increasing accessibility and equity; (ii) allowing the rationalisation of any additional testing and (iii) promoting an early approach, as each of the 10 child psychiatry sessions also had a therapeutic objective, since they were designed according to the D.I.R. model.

This model favours the organisation of team staff, (i) preventing duplicate consultations; (ii) making complementarity possible and (iii) promoting the links with community structures, which are very relevant for medium- and long-term follow-up.

This study was aimed at the assessment of the innovation provided by the recent development of this unique National Health Service structure, the CEBC and of this model for the follow-up of patients aged 0-3 with suspected ASD. The model involves the experience of departments with renowned merit and may bring important gains at short and medium term, also helping to rethink the local structures required to follow these children.[34] We believe that it will become a proven valid model that will be used by different other departments with initial technical supervision.

CONTRIBUTION OF THE AUTHORS

CMH, PCS, DC, MJN: Significant direct intellectual contribution to design and development of the manuscript, data analysis and final version approval.

JMR, MTM: Significant direct intellectual contribution to design and development of the manuscript, data analysis and final version approval.

BPF, IS, LC, MPG, MM, MJP, PL, PS, RR, SC, SAP, SP, SA: Significant intellectual contribution in draft review and final version approval.

HUMAN AND ANIMAL PROTECTION

The authors declare that this project complied with the regulations that were established by the Ethics and Clinical Research Committee, according to the 2013 update of the Helsinki Declaration of the World Medical Association.

DATA CONFIDENTIALITY

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

CONFLICTS OF INTEREST

The authors declare that there were no conflicts of interest in writing this manuscript.

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