Aicardi’s Syndrome is a severe autosomal recessive neurodegenerative encephalopathy identified in 1965 by Jean Aicardi and described in literature since 1984 by Aicardi and Goutières [Aicardi, 2005]. It is a rare genetic disease, 853 case are estimated in the US and over 4000 worldwide [Kroner et al., 2008]; it is not inherited and happens by a new genetic change (de novo mutation) [Margaret et al., 2006].

The diagnosis is based on: microencephaly, spasticity and severe mental delay. It affects only females because of early embryonic lethality in males. A significant number of females with Aicardi syndrome are of normal birth and develop normally until three months of age when infantile seizures begin. Psychomotor retardation is usually severe, neuromotor retardation is commonly present with lack of motor and language skills. Literature reports only few information about the dentofacial features of the syndrome. We present the case of a patient, by describing the dental and facial characteristics with focus on dental prevention in order to avoid dental pain and the risks connected to general anaesthesia, and ultimately for improving the quality of life.

Key words: Aicardi Syndrome, main features, clinical findings, dental prevention

Introduction

Aicardi’s Syndrome is a severe autosomal recessive neurodegenerative encephalopathy identified in 1965 by Jean Aicardi and described in literature since 1984 by Aicardi and Goutières [Aicardi, 2005]. It is a rare genetic disease, 853 case are estimated in the US and over 4000 worldwide [Kroner et al., 2008]; it is not inherited and happens by a new genetic change (de novo mutation) [Margaret et al., 2006].

The diagnosis is based on: microencephaly, spasticity, severe mental delay and retinal lesions [Abdel-Salam et al., 2004; Aicardi et al. 1965; Lanzi et al., 2002; Le Garrec et al., 2005].

The disease is characterised by agenesis of the corpus callosum, either partial or complete, associated with cerebral palsy, epileptic-like symptoms, and infantile spasm (a form of seizure). It only affects females because of early embryonic lethality in males and, in very rare cases, males with Klinefelter syndrome (47, XXY).

It has been hypothesised X-chromosome linked anomalies, but genetic researches have not yet identified the genes involved in the syndrome. Almost all of the cases are new mutations so sibling and other family members do not carry the defective gene. The prevalence is extremely low [Aicardi, 1998; Crow, 2002; Fishbach, 2002].

A significant number of Aicardi syndrome girls are of normal birth and of normal development until three months of age when infantile spasms begin. Infantile spasms are the first symptoms that urge the clinicians to prescribe further investigations [Prats Viñas, 2005]. Cerebral malformations like polymicrogyria, grey matter heterotopias, intraventricular cysts, porencephalic cysts and chorid plexus papillomas coexist at times. Occasional finding are: abnormalities of ribs and spine, cleft palate and ocular anomalies [Aicardi, 2005; Aicardi, 1998; Allison et al., 2004; Banerjee et al., 2006; Bastos et Borges, 2001; Melbourne-Chambers et al., 2007; Rosser et al., 2001] (Table 1). Psychomotor retardation is usually severe; neuromotor retardation is commonly present with lack of motor and language skills [Rosser et al., 2002; Grosso et al., 2007].

American researchers have recently defined the typical facial features of the affected females: prominent maxilla, upturned nasal tip, decreased angle of the nasal bridge, sparse lateral eyebrows [Sutton et al., 2005].

Case report

TB, born in 1998, showed agenesis of corpus callosum since the 6th week of pregnancy. After birth ocular anomalies (bilateral ocular coloboma, chorioretinal lacunae), cerebral abnormalities (dysmorphic and enlarged ventricles, asymmetry of the emispheres) and vertebral anomalies (butterfly shaped) were detected. Mental delay is severe, general hypotonia and motor delay make walking unlikely to happen.

She came to the Unit for Special Care Patients of the Dental School of the University of Bologna (Italy) at 4 years of age for dental evaluation because parents referred...
frequent episodes of acute pain.

The patient exhibited normal deciduous dentition (Fig. 1) and showed occlusal decays on all molars and deep caries of the front upper and lower teeth. The diagnosis was ECC (Early Childhood Caries). She had drug-resistant seizures, and the administration of various medications made the gingival aspect typical of a patients with antiepileptic drugs-induced overgrowth and inflammation (Fig. 2, 3.)

Tongue position was very low and swallowing reflex and chewing were absent. She was fed a soft and semiliquid diet.

The malocclusion was characterised by severe open bite (only the four second deciduous molars are in occlusion) and spacing of the upper and lower incisors because of the low tongue position, narrow palate worsened by a severe gingival overgrowth, lateral left scissorbite (Fig. 4).

Because of the severe psychomotor impairment, she is on a wheelchair in a tilted-back position because she cannot bear her own skull weight due to general muscle hypotonia. The posture of the head is hyperextended all the time and the mouth is always open, the open bite is therefore worsened by the permanent mouth breathing and the muscle hypotonia. Cooperation is very low and not enough to execute correct X-rays to perform a skeletal evaluation and a proper orthodontic treatment plan.

Her disruptive behaviour makes even conservative dental procedures and hygiene very difficult to perform. She was prescribed antibiotic therapy (amoxicillin 50mg/kg/die for 5 days) to ease the infection, and then she underwent pulpectomy of the upper incisors, and fillings of decayed pits of the deciduous molars.

The mother (the only caregivers accepted by the girl) was instructed on brushing technique with the aid of a mouth prop, and was given dietary advices to avoid/reduce the caries risk.

Periodic dental checkups were scheduled (every 3-6 months) to perform professional hygiene procedures.

Home hygiene maintenance is not easily performed by the parents because of the girl's behaviour and because parents are not comfortable in using a mouth prop to keep the arches open.

In November 2006 the patient came to our attention and showed a severe erosive mucositis in the palatal area, secondary caries of the molar pits of the primary teeth and a bite lesion of the cheek at the level of the scissor bite (Fig. 5).

The oral smear revealed a candida infection that was treated with Mycostatin per os for 7 days and conservative treatment were scheduled. We decided to treat the cheek lesion with an orthodontic appliance in order to correct the lingual inclination of the lower left molar.

Some international papers have investigated the problems that Aicardi Syndrome girls may have in case of general anaesthesia. In fact severe generalized hypotrophia of subcutaneous tissues cause a difficult
venous access in the limbs, and severe vertebral malformation makes a caudal block impossible [Iacobucci et al., 2003]. Tongue hypotonia and consequently macroglossia can complicate the intubation procedures [Gooden et al., 2005].

Discussion

Patients affected by Aicardi Syndrome show a severe neurodegenerative encephalopathy and subsequent motor and mental delay. The syndrome induces cerebral atrophy, intracerebral calcification and developmental arrest. The main features include a triad of infantile spasms, agenesis of corpus callosum and chorioretinal lacunae. Diagnosis requires the presence of progressive encephalopathy with clinical onset shortly after birth. The typical facial dismorphic features are: microphthalmia, prominent maxilla, upturned nasal tip, decreased angle of the nasal bridge and sparse lateral eyebrows.

Behavioural management of these patients is capital but very difficult because of the severe mental impairment and lack of cooperation. Poor oral health is common in disabled patients, especially those with severe mental delay, because of the inability to perform even simple activities. Feeding and chewing ability are restricted to soft food and liquids. Hypotonia of the masticatory muscles, lip incompetence, low mobility of the tongue and drowsiness reduce even the scarce “self-cleaning” action of saliva.

Frequent intake of sweets to provide an adequate calories intake, sugar added drugs administration, drug-induced gingival overgrowth due to antiepileptic medications, and inadequate oral hygiene can threaten the oral health of these subjects.

Dental therapy under conscious sedation may help the operators to perform safe therapies bypassing the lack of collaboration and preventing the risks connected to general anaesthesia.

Conclusion

Dental checkups and family counselling focused on primary oral prevention are capital and they should be part of the periodic medical follow up. On the contrary, dental intervention is often required in case of emergency and pain episodes when primary prevention is no longer feasible and performing invasive dental procedures is complicated.

Prevention of oral diseases is the key of success for special care patients. It could reduce the onset of dental caries and gingival disease, and therefore the need of dental therapies (especially under general anaesthesia) in order to avoid medical complications, pain experience and improve the quality of life of the girls affected by Aicardi Syndrome.

References


