Callosal agenesis, chorioretinal lacunae, absence of infantile spasms, and normal development: Aicardi syndrome without epilepsy?

Jose Prats Viñas and coworkers’ (p 419) report in this issue is of great interest, first because it raises the problem of the definition and limits of Aicardi syndrome but also, accepting as I do that it belongs to the syndrome, it suggests a much wider spectrum of expression.

The features of the syndrome, as initially described, included the triad of agenesis of the corpus callosum, spasms in flexion, and chorioretinal lacunae, the latter being especially characteristic and generally regarded as pathognomonic. Vertebrocostal anomalies were also reported but not as a constant feature. More recently, additional features have been recognized, including the presence of periventricular heterotopias of grey matter, intracranial cysts (especially inter-hemispheric and around the third ventricle), cysts and/or papillomas in the choroid plexus, and gross ventricular asymmetry, which contribute to highly suggestive gestalt on imaging. These striking abnormalities, however, are probably not responsible for seizures and learning disability* and the most important anomaly is likely to be migration disturbances, which may be very difficult to detect even with learning disability. Perhaps, in the patient of Prats Viñas et al. they were mild, which could also explain the benign course. Whether all the constituents of the triad are necessary for diagnosis has been previously questioned.1 Cases with a complete corpus callosum and cases without spasms have been described. I have seen two female children with normal fundi. The absence of any reliable marker (biochemical or genetic) has not allowed a firm answer. Learning disability has been considered inevitable. However, cases with milder manifestations or with controllable seizures are on record, usually in association with with some atypical features. Although the frequency of such cases is unknown, and likely to be low, the possibility should be considered before giving a gloomy prognosis. The final answer will have to await the discovery of a reliable marker.

Jean Aicardi

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Reference


Influence of supine sleep positioning on early motor milestone acquisition

In this issue Majnemer and Barr (p 370) bring to our attention two groups of 4 and 6-month-old infants, who had spent much of their time supine. As the authors describe some aspects of their immediate early development, I find myself being repeatedly reminded of other associated and longer-term aspects of position-related development. In their discussion the authors rightly remind us that ‘infant motor development involves a dynamic interplay between intrinsic maturation of the musculoskeletal and nervous systems and extrinsic experiences’. But what about genetic bias?

Trying to improve diagnosis of cerebral palsy very early in life at The Newcomen Centre, Guy’s Hospital, London during the 1960s we were faced with the comparatively wide variability in normal locomotor development. It was well known that there were many normal children who did not conform (some ‘delayed’ and others ‘exceptional’).

Yet surprisingly the majority of the population were allowed to set the ‘norms’ for all motor development. Statistically there was more than one normal locomotor timescale and several locomotor sequences.

Our population was mainly in south-east London, and Robson went on to define five locomotor populations and record their incidence.1,2 Interestingly two groups, both supine developers and making up 16% of our population, appeared to have a dominantly inherited trait. Were we studying an exceptional population? I doubt it. We all recognize these developmental disparities and others have reported their local incidence.3 They also exhibited a marked resistance to being lain prone (leading to early prone-to-supine rolling; also noted by Majnemer and Barr in their population).

Longitudinal study4 has demonstrated that, for children placed in their appropriate gross motor developmental group, meaningful correlations can be shown between one gross locomotor ‘skill’ and another, and that these data can help clinicians and reassure parents. Not all children with delay slot neatly into a group, but many of those causing parental concern do. For me, that is good enough.

David Scutt

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References


*US usage: mental retardation.