Microphthalmia with linear skin defects syndrome (MIDAS)

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DESCRIPTION

Congenital skin atrophic lesions are infrequent and generally benign but may be associated with underlying genetic conditions.

We describe the case of a newborn girl, second offspring of a non-consanguineous healthy couple, that was born full term after an uneventful pregnancy, with an apgar index of 9/10 and normal weight-for-age. On examination, she had left eye (LE) microphthalmia, right eye anophthalmia, skin lesions of dermal aplasia involving eyes and cheeks (figure 1) and upper limbs (figure 2), following Blaschko's lines, right pit and anterior anus.

Chest X-ray, renal, abdominal and transfontanellar ultrasound imaging were unremarkable. Ophthalmic evaluation also confirmed LE corneal opacification and neovascularisation. Orbital magnetic resonance imaging (MRI) showed absence of LE lens and right optic nerve hypoplasia. Cerebral MRI revealed hypoplasia of the corpus callosum, septum pellucidum agenesis and hypoplasia right opaque bulb. Brainstem Auditory Evoked Potential revealed bilateral hypoacusis with middle ear effusion.

There were no structural cardiac anomalies, but 24 hours ECG monitoring showed ventricular extrasystoles, bigeminism, trigeminism and quadrigeminism.

At the age of 20 days, she developed supraventricular tachycardia that converted to normal sinus rhythm after adenosine. Therapy with propranolol was started, after which no further episodes occurred.

Figure 1 Lesions of dermal aplasia involving eyes and cheeks.

Figure 2 Lesions of dermal aplasia following Blaschko's lines.

Karyotype analysis revealed a complex mosaic karyotype, with two abnormal cell lines: mos 46,X,der(X)del(X)(p22.2)dup(X)(p21.1p22.2)[24]/45,X[6]dn. Karyotype of both parents was normal, confirming these as de novo alterations.

Array comparative genomic hybridisation (CGH) revealed a Xp terminal deletion: (arr[GRCh37] Xp22.33p22.2 (296520_12432327)x1) encompassing HCCS gene. In addition, a 22.78 Mb Xp22.2p21.1 duplication involves 79 OMIM genes in other syndromic regions was also detected.

Microphthalmia with linear skin defects syndrome (MIDAS) (MIM 309801) is a rare syndrome with an X-linked dominant transmission pattern, with male lethality.¹ It is associated with deletions or unbalanced translocations in the short arm of X-chromosome (Xp22.3).² Most cases are de novo, but familial recurrence has been described, with no clear genotype–phenotype correlations.³

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The most consistent clinical features of MIDAS are microphthalmia and/or anophthalmia, sclerocornea and linear skin defects with dermal aplasia limited to face and neck, which were all present in this case and raised the clinical suspicion before the final diagnosis. Nervous system and cardiac anomalies, development delay, diaphragmatic hernia and genitourinary tract abnormalities, hearing loss and anal atresia with ectopic anus and fistula can also be present.¹⁻³

The use of array CGH as the first laboratorial approach in the investigation of a newborn with a predicted diagnosis of MIDAS syndrome can confirm the diagnosis and may identify

unsuspected copy number variants.³ The phenotype associated with the deletion found, can range from no clinical signs to in utero lethality, depending not only on somatic mosaicism, but also on cell selection mechanisms.³ The contribution of the duplication (Xp22.2p21.1) to the clinical manifestations here is unknown.

The patient is currently under multidisciplinary follow-up. An LE prosthesis was implanted at 3 months. Neuropaediatric evaluation revealed upper and lower limbs hypotonia, with no other changes in psychomotor development.

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Learning points

- MIDAS is characterised by microphthalmia and/or anophthalmia, sclerocornea and linear skin defects with dermal aplasia limited to face and neck, which were all present in this case and raised the clinical suspicion.
- The use of comparative genomic hybridisation as the first laboratorial approach in the investigation of a newborn girl with a predicted diagnosis of MIDAS syndrome is recommended.
- Transmission occurs in X-dominant pattern, with male lethality. Most cases are de novo.

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