



ORIGINAL ARTICLE

Medical management of children with achondroplasia: Evaluation of an Australasian cohort aged 0–5 years

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Aims: Achondroplasia is the most common form of osteochondrodysplasia and is associated with a number of life-threatening complications. The complexity of the condition led to the development of Health Supervision Guidelines published by the American Academy of Pediatrics in 1995 and revised in 2005. There remains limited population-based information on utilisation of medical and therapy services for children with achondroplasia. Increased information regarding use of these services will assist in future service development.

Methods: Data regarding frequency and timing of medical and allied health consultations, investigations and interventions were collected from 53 Australasian families via questionnaire, based on recommendations of the Health Supervision Guidelines, an expert reference group and literature review.

Results: Rates varied with age for medical consultations (geneticist, paediatric rehabilitation physician/paediatrician, respiratory physician, orthopaedic consultant, neurologist, neurosurgeon), medical investigations (sleep study, magnetic resonance imaging/computed tomography), operative procedures (brain-stem decompression, tonsillectomy/adenoidectomy, shunt insertion, shunt revision and insertion of grommets) and allied health consultations (physiotherapist, occupational therapist, speech pathologist, dietician and orthotist).

Conclusions: Access to geneticists and paediatricians within the first year is high as recommended by the 2005 American Academy of Pediatrics guidelines. Utilisation of craniocervical magnetic resonance imaging/computed tomography, polysomnography studies and formal speech review appears low, reflecting more emphasis on clinical monitoring for cervical cord compression and disordered sleep breathing as well as possible difficulties in accessing services for polysomnography and speech pathology. Grommet insertion, tonsillectomy/adenoidectomy and cervicomedullary decompression rates are similar to results reported previously. Over half of the children accessed physiotherapy and/or occupational therapy services, warranting consideration of these professionals in future guideline recommendations.

Key words: achondroplasia; allied health personnel; management, medical practice.

Achondroplasia is the most common form of skeletal dysplasia with an estimated incidence between 0.36 and 0.6 per 10 000 live births.^{1–6} Achondroplasia is associated with many common morbidities involving the neurological, musculoskeletal and cardiorespiratory systems which in some children lead to potentially life-threatening medical complications. However, while this results in an increased rate of mortality at all ages,⁷ starting with estimates of 2.7 to 7.5% in infancy,^{7–9} most individuals with achondroplasia are of average intelligence and are inde-

pendent in social and employment aspects of life.^{4,10} The goal for the multidisciplinary team therefore is to have in place adequate monitoring and management systems to enable early identification and treatment of emergent problems.

Many medical complications associated with achondroplasia relate to the disproportion existing between the growth in endochondral bones versus other tissues/organs.⁶ Individuals with achondroplasia may have a small foramen magnum with up to 35% demonstrating cervicomedullary compression on imaging.¹¹ In a very small proportion of infants, resultant cervicocervical cord compression can lead to quadriplegia, hypopnoea or sudden infant death.^{7,9,11} Other concerns include hydrocephalus, with 10% of children reported to have ventricular shunts in place by teenage years.¹² Midface hypoplasia, relative adenotonsillar hypertrophy, potential posterior cranial fossa compression and stenosis of the upper airway can contribute to

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What is already known on this topic

- 1 Achondroplasia is a complex medical condition with a number of life-threatening complications impacting upon the neurological, musculoskeletal and cardiorespiratory systems.
- 2 The Health Supervision Guidelines for children with achondroplasia published by the American Academy of Pediatrics (AAP) offer clinicians clear suggestions from a North American perspective for the multidisciplinary assessment and management of this group of patients.
- 3 Despite the known complexity of the condition and publication of treatment recommendations, there is still little information regarding the multidisciplinary medical and therapy practices and service usage for children with achondroplasia.

What this paper adds

- 1 The proportion of Australasian children accessing geneticists and paediatricians within the first 12 months is approaching the 2005 North American recommendations for children with achondroplasia, while a significant proportion of Australasian children are still not accessing speech and language pathology services.
- 2 While utilisation of magnetic resonance imaging/computed tomography and polysomnography studies appears low, the number of children undergoing craniocervical decompression (cervicomedullary decompression) and tonsillectomy/adenoidectomy is similar to results reported in previous studies suggesting that there may be other non-invasive assessments that clinicians are using to accurately determine risk and need for intervention.
- 3 Despite there being no recommendation in the 2005 AAP guidelines, over 75% of Australasian children with achondroplasia accessed physiotherapy services in the first three years, and inclusion of these professionals as part of a designated multidisciplinary team should be considered in future service planning and development.

sleep-disordered breathing, believed to affect up to 85% of children with achondroplasia^{13–18} or sudden infant death.^{6,11,15,19} Altered midface anatomy, narrow nasal cavities and shortened Eustachian tubes lead to an increased frequency of ear infections and hearing loss^{2,3,12,15} with approximately 50% of individuals undergoing insertion of grommets during their lifetime.^{12,14,20} Narrowing of the interpedicular distances and vertebral wedging at the thoracolumbar region contribute to symptomatic spinal stenosis, more common in adults, with up to 80% of 50-year-olds presenting with leg pain and neurological signs.¹² Other recognised features include a thoracolumbar gibbus, present in 95% of newborns,²¹ tibial bowing and delayed motor, speech and feeding development.^{22–24}

Early management with interventions aimed at preventing later complications is important for reducing morbidity and mortality rates for individuals with achondroplasia.^{4,7,9,12} Recognition of the type and timing of common complications led to the development of clinical guidelines for 'Health Supervision of Children with Achondroplasia' by the American Academy of Paediatrics (AAP).^{2,4} These guidelines assist clinicians in identifying children at risk of life-threatening complications with clear suggestions on management. For example, early management of infants with achondroplasia in Australia includes reclined handling commencing in the perinatal period and continuing until the child can hold its head and trunk unsupported. This programme, recommended in the 1988 paper of Hall and both the 1995 and 2005 AAP guidelines, has been implemented by Australian centres since 1988. While Hunter *et al.*¹² offered valuable data on the rates and ages of occurrence for several complications and medical interventions, there remains limited population-based information on patterns of utilisation of medical and therapy services in Australia since publication of the AAP Guidelines in 1995 and 2005.

This study reports Australasian service statistics on the type and timing of multidisciplinary medical and allied health assess-

ments and interventions provided for children with achondroplasia born since 1997. A clearer understanding of the current utilisation of medical and therapy services will help to drive and direct future service development for this group and enhance outcomes for individuals with achondroplasia.

Methods

This is a cohort study of children with achondroplasia born in Australasia (Australia and New Zealand) from January 1997 to December 2007. Ethical approval was obtained from human research ethics committees of participating hospitals in each state.

Participants

Children were recruited through bone dysplasia and clinical genetics clinics at major hospitals in all six Australian states and through the Short Statured People's Association in Australia and the Little People's Association in New Zealand. Parents were contacted by mail and invited to participate. Individuals were eligible if they had a diagnosis of achondroplasia and were aged less than 5 years at the time of enrolment. Children with concomitant medical problems such as epilepsy or cerebral palsy were excluded.

Medical history questionnaire

A questionnaire was developed to gather information about the frequency and timing of recommended medical and allied health consultations, investigations and interventions. Content validity of the questionnaire was established by three methods.

- 1 Recommendations were sought from a multidisciplinary reference group with substantial experience in the care of children with achondroplasia. This group identified 11 different

health professionals and seven specific medical procedures commonly associated with long-term care in this population.

- The questionnaire also included recommendations made in the guidelines for 'Health Supervision for children with Achondroplasia' published by the Committee on Genetics of the American Academy of Paediatrics (1995 and 2005). In summary, these recommendations included: (i) genetic counselling within the first 12 months of life; (ii) consultation with a paediatric physician with expertise concerning achondroplasia to monitor general health; (iii) referral as necessary to a neurosurgeon or other physician skilled and experienced in the care and treatment of neurologic problems in children with achondroplasia; (iv) formal speech evaluation no later than 2 years of age; (v) assessment for craniocervical junction risks, including careful neurologic history and examination and neuro-imaging (computed tomography (CT) or magnetic resonance imaging (MRI)); and (vi) polysomnography (sleep study). For more information, a full copy of the guidelines can be accessed free online at: <http://pediatrics.aappublications.org/cgi/reprint/116/3/771>.
- Finally, a literature review was conducted to identify any further morbidities and/or frequently reported or recommended medical and allied health assessments or interventions, such as tonsillectomy, grommet insertion, speech problems, ventricular shunting and cervicomedullary decompression (CMD).^{12,14,16,20,21}

Procedure

When a child was enrolled, parents were asked to complete the questionnaire with information about their child's medical, surgical and allied health consultations, investigations and interventions to date. Families then received the questionnaire every 3 months up to their child's 5th birthday (or the end of data collection, if this came first) to record the consultations, investigations and interventions that had occurred in the preceding 3-month period. Parents were asked to include services received from all sources, including tertiary hospitals, primary health-care facilities, non-government community and/or private services.

Statistical analysis

Descriptive statistics were calculated, including the frequency and percentage of each consultation, investigation and intervention at each age. These results were compared with recommendations made in the American Health Supervision Guidelines (1995:2005).

Results

Seventy-four Australasian children aged 0 years to 5 years 0 months with achondroplasia were identified during the study period. Fifty-five families (74%) enrolled and 53 families (72%) completed questionnaires over at least 3 years. Two families were lost to follow-up. Twenty-seven participants were boys. Twenty-seven children attended services through specialist multidisciplinary clinics co-ordinated specifically for children with bone dysplasias, and the remaining 26 attended tertiary paediatric hospitals providing services for children with complex conditions. Enrolment age ranged from 1 to 60 months with a mean age of 21 months. At the time of reporting, data were available for 41 children up to their 5th birthday, for 50 children up to their 4th birthday and for 53 children up to their 3rd birthday.

Medical consultations

The majority of the Australasian-born children with achondroplasia studied were reviewed in the first 12 months by a geneticist and/or specialist in paediatric rehabilitation/paediatrician as recommended in the Health Supervision Guidelines 2005 (Table 1). Neurologists (20%; 11/53) and neurosurgeons (28.3%; 15/53) were the least frequently seen medical specialists over the first 3 years.

Investigations

In this cohort, 64% (32/53) of children had formal polysomnography studies by age 3, with over one third (20/53) of these studies occurring within the first 12 months (Table 2). Twenty-two children (41.5%) were reported as having MRI or CT studies by age 3.

Table 1 Number (%) of recruited Australasian children with achondroplasia ($n = 53$) receiving consultations by medical subspecialists in the first 3 years of life

Medical subspecialist	0–1 years Total (%)	1–2 years Total (%)	2–3 years Total (%)	Not seen by age 3 Total (%)
Geneticist	44 (83.0)	30 (56.6)	20 (37.7)	6 (11.3)
Paediatrician	46 (86.8)	34 (64.1)	28 (52.8)	6 (11.3)
Respiratory physician	15 (28.3)	18 (34.0)	16 (30.2)	28 (52.8)
Neurologist	7 (13.2)	9 (16.9)	5 (9.4)	42 (79.2)
Orthopaedic surgeon	10 (18.9)	17 (32.1)	16 (30.2)	31 (58.5)
Neurosurgeon	10 (18.9)	13 (24.5)	8 (15.1)	38 (71.7)

Total (%), combined number of new and follow-up consultations (and that total expressed as a percentage of children recruited at that age).

Table 2 Number (%) of recruited Australasian-born children with achondroplasia undergoing medical investigations or surgical procedures by 5 years of age

	Group (n = 53)				Group (n = 50)		Group (n = 41)	
	0–1 years Total (%)	1–2 years Total (%)	2–3 years Total (%)	Not seen by age 3 (n/%)	3–4 years Total (%)	Not seen by age 4 (n/%)	4–5 years Total (%)	Not seen by age 5 (n/%)
Polysomnography (sleep study)	20 (37.7)	22 (41.5)	15 (28.3)	21 (39.6)	13 (26)	16 (32)	9 (21.9)	14 (34.1)
MRI/CT	16 (30.2)	14 (26.4)	11 (20.8)	31 (58.5)	8 (16)	27 (54)	7 (17.1)	21 (51.2)
Grommets	9 (16.9)	17 (32.1)	19 (35.8)	23 (43.4)	8 (16)	20 (40)	10 (24.4)	13 (31.7)
Tonsillectomy/adenoidectomy	5 (9.4)	10 (18.9)	10 (18.9)	32 (60.4)	6 (12)	26 (52)	3 (7.3)	19 (46.3)
Brain-stem decompression	4 (7.5)	3 (5.7)	1 (1.9)	45 (84.9)	0 (0)	42 (84)	0 (0)	34 (82.9)
Shunt inserted	1 (1.9)	0 (0)	1 (1.9)	52 (98.1)	0 (0)	49 (98)	0 (0)	40 (97.6)
Shunt reinserted	0 (0)	0 (0)	1 (1.9)	52 (98.1)	1 (2)	49 (98)	0 (0)	40 (97.6)

CT, computed tomography; MRI, magnetic resonance imaging; total (%), combined number of new and follow-up investigations or procedures (and that total expressed as a percentage of children recruited at that age).

Operative procedures

Insertion of grommets was the most common operative procedure for children under 3 years, with over 50% of children (30/53) undergoing this procedure (Table 2). Forty per cent (21/53) of children had removal of tonsils and/or adenoids in the first 3 years with just over half (53.6%; 22/41) reporting this by 5 years. Lumboperitoneal or ventriculoperitoneal shunt insertion and/or revision was the least common procedure reported, with only one child requiring this surgery by age 5.

Allied health consultations

Physiotherapists were the most frequently seen allied health professionals during the first 3 years of life, while occupational therapists and speech pathologists were the most commonly consulted allied health professionals during the fourth and fifth years (Table 3). Despite known problems with early language development, frequency of referral to a speech and language pathologist was low, with only 18.8% (10/53) accessing a speech and language pathologist in the first 12 months. While contact increased, numbers still only reached 58.5% (31/53) by age 3 and 61% (25/41) by age 5. Dieticians (21%; 11/53) and orthotists (7.5%; 4/53) were the least frequently consulted allied health practitioners.

Discussion

This study provides a summary of medical and allied health consultations, investigations and interventions received by Australasian children with achondroplasia aged 0–5 years during the period 1997–2010. Data were collected from all consenting families within the Australasian cohort. Using the identified population to calculate incidence of achondroplasia results in an estimated incidence rate of 0.26 per 10 000 births which is similar to that reported by Waller *et al.*,⁵ suggesting that the majority of children were identified. While Health Supervision

Guidelines for Children with achondroplasia published by the American Academy of Pediatrics in 1995 and revised in 2005 are not purported as a definitive programme of treatment^(2,4), they do offer clear recommendations for multidisciplinary assessment and management and suggested referral and intervention.

The highest level of compliance with the published guidelines for medical consultation services was achieved by geneticists and paediatric rehabilitation physicians/paediatricians within the first year of life. This very high level of access to recommended frontline subspecialists is a major benefit of tailored services for children with achondroplasia in Australasia, where the common model for bone dysplasia clinics includes paediatric rehabilitation physicians/paediatricians and geneticists with referral to other medical subspecialists as indicated.

Compliance with recommended investigations, including polysomnography (sleep studies) and MRI/CT scans was low. Parents reported that around one third of children in this group had MRI/CT (30%). However, Australian best practice is to monitor clinically at 3 months intervals in the first 18 months, plotting head growth on published achondroplasia charts, and monitor ventricular size by ultrasonography via the anterior fontanelle rather than MRI/CT. The office neurologic examination also includes assessment of tone, reflexes, clonus and plantar response reserving CT/MRI for patients where it is clinically indicated (Professional Advisory Committee, Short Statured People of Australia, unpubl. data, 2010). Similarly, polysomnography for sleep-disordered breathing was achieved for only 38% in the first 12 months. However, by age 5, only one third of children had not had a formal polysomnography study. It is not known whether this was because referral was not clinically warranted, although previous Australian studies have established the value of referral of all infants and children to a respiratory paediatrician/paediatric sleep medicine service.¹⁷ Anecdotally, there has been difficulty in accessing these investigations particularly for geographic or service capacity reasons. A number of groups advocate that MRI/CT and polysomnography be undertaken as part of routine surveillance in the neonatal period or at time of diagnosis.^{4,7,9} Given that there has been

Table 3 Number (%) of recruited Australasian-born children with achondroplasia attending allied health sessions in the first 5 years of life

	0–1 years (n = 53) n (%)	1–2 years (n = 53) n (%)	2–3 years (n = 53) n (%)	Not seen by age 3 n (%)	3–4 years (n = 50) n (%)	Not seen by age 4 n (%)	4–5 years (n = 41) n (%)	Not seen by age 5 n (%)
<i>Physiotherapist</i>								
1–4 visits	18 (52.9)	13 (54.2)	17 (68)	NA	17 (94.4)	NA	8 (88.9)	NA
5–10 visits	11 (32.4)	4 (16.7)	3 (12)	NA	0 (0)	NA	0 (0)	NA
>10 visits	5 (14.7)	7 (29.2)	5 (20)	NA	1 (5.6)	NA	1 (11.1)	NA
Total (%)	34 (64.1)	24 (45.3)	25 (47.2)	13 (24.5)	18 (36)	13 (26)	9 (22)	12 (29.3)
<i>Occupational therapist</i>								
1–4 visits	10 (71.4)	9 (64.3)	12 (66.6)	NA	21 (95.5)	NA	11 (91.7)	NA
5–10 visits	3 (21.4)	2 (14.3)	1 (5.6)	NA	0 (0)	NA	0 (0)	NA
>10 visits	1 (7.2)	3 (21.4)	5 (27.8)	NA	1 (4.5)	NA	1 (8.3)	NA
Total	14 (26.4)	14 (26.4)	18 (33.9)	26 (49.1)	22 (44)	15 (30)	12 (29.3)	13 (31.7)
<i>Speech pathologist</i>								
1–4 visits	5 (50)	16 (84.2)	14 (66.7)	NA	11 (64.7)	NA	6 (54.5)	NA
5–10 visits	4 (40)	2 (10.5)	4 (19.0)	NA	2 (11.8)	NA	2 (18.2)	NA
>10 visits	1 (10)	1 (52.6)	3 (14.3)	NA	4 (23.5)	NA	3 (27.3)	NA
Total	10 (18.9)	19 (35.8)	21 (39.6)	22 (41.5)	17 (34)	17 (34)	11 (26.8)	16 (39)
<i>Dietician</i>								
1–4 visits	5 (83.3)	6 (100)	4 (100)	NA	4 (100)	NA	3 (100)	NA
5–10 visits	0 (0)	0 (0)	0 (0)	NA	0 (0)	NA	0 (0)	NA
>10 visits	1 (16.7)	0 (0)	0 (0)	NA	0 (0)	NA	0 (0)	NA
Total	6 (11.3)	6 (11.3)	4 (7.5)	42 (79.2)	4 (8)	38 (76)	3 (7)	29 (71)
<i>Orthotist</i>								
1–4 visits	0 (0)	2 (100)	3 (100)	NA	4 (100)	NA	2 (100)	NA
5–10 visits	0 (0)	0 (0)	0 (0)	NA	0 (0)	NA	0 (0)	NA
>10 visits	0 (0)	0 (0)	0 (0)	NA	0 (0)	NA	0 (0)	NA
Total	0 (0)	2 (3.8)	3 (5.7)	49 (92.5)	4 (8)	44 (88)	2 (4.9)	35 (85)

(%), percentage of total children accessing individual allied health service; NA, not applicable; total (%), number children in cohort accessing individual allied health service; (expressed as a percentage of total children recruited at that age.

careful attention to advice about early handling and a programme of systematic clinical monitoring for neurologic complications, the risk for increased mortality rates should be low. On the other hand, the increasing accessibility of polysomnography could assist clinicians in identifying infants and young children at high risk of sudden infant death related to stenosis at the craniocervical junction with compressive damage to the respiratory control centres.^{9,17,25} No children recruited to the study cohort died during the study period.

Medical and surgical interventions were within the range of intervention rates reported for previous cohorts. In our cohort, seven of the 53 children (13.2%) reported CMD within the first 2 years, and eight out of 50 (16%) reported CMD by 4 years. Pauli *et al.* in 1984⁹ noted a similar rate with 10 out of 75 children (13.3%) requiring CMD, mostly within the first 2 years. Lower rates were reported by Hunter *et al.* in 1998¹² (6.8% by 4 years) and in the most recent report by King *et al.* in 2009²⁶ (13 of 162 (8%) children since 1988 with 11 (6.7%) within the first 2 years). Ho *et al.* in 2004²⁷ reporting from a neurosurgical service recorded a notably higher rate of 28% of children by 4 years. With the exception of the cohort of Ho *et al.*, this suggests that irrespective of potential differences in monitoring procedures, CMD rates for children under 6 years

have stayed fairly consistent at between 6 and 16% internationally over the last 25 years.

While ventriculomegaly and excessive extra-axial fluid are seen in children with achondroplasia, it is generally a communicating hydrocephalus and does not routinely require shunting, unless associated with a rapid increase in head size or symptoms of increased pressure.⁶ Only one child in our cohort reported placement of a shunt, which is consistent to the next most recent data by King *et al.* in 2009²⁶ who reported one child with a shunt insertion over the last 20 years. A higher rate was reported by the earlier study by Hunter *et al.* in 1998¹² with 12 children (6.48%) in their cohort requiring shunt insertion before the age of 5. This suggests that increased understanding of the type of hydrocephalus commonly presenting in achondroplasia, together with the availability of standard growth charts, and non-invasive ultrasound monitoring has led to a decrease in intervention rates.

Utilisation of allied health services by our cohort provides a previously undocumented statistic supporting the need for multidisciplinary management of children with achondroplasia. Physiotherapists were the most frequently consulted allied health professionals during the first 3 years. Most input occurred in the first year (64.1%).

The guideline recommendation for speech pathology review within the first 2 years reflects the frequency of midface hypoplasia, short Eustachian tubes and relative adenotonsillar hypertrophy that contribute to high otitis media rates,¹⁴ associated hearing loss and, thus, communication skill delays reported in this group.^{22,23} When the total numbers of children requiring insertion of grommets in our cohort were reviewed by year and compared with figures reported by Hunter *et al.*,¹² the numbers were found to be very similar with both studies reporting an almost identical percentage of children not having undergone insertion of grommets by age 5 (31.7% vs. 31.5%). The numbers of children accessing speech pathology, a guideline recommendation, were low with 60% of children being reviewed by a speech pathologist by 5 years and just under half occurring before 2 years of age. Our data indicate a significant increase compared to previous cohorts, for example Hunter *et al.*'s study¹² reported no speech pathology input prior to age 2 and only 4.6% compliance (eight out of 174) by age 5. This suggests a greater understanding of the need to monitor and manage delays in communication and feeding skills.^{22,23} It is not possible to ascertain if the gap in uptake in this study reflected reduced referral rates by service providers, poor attendance by families or good compliance in an environment of service limitations or access difficulties. The need for occupational therapy was similar to speech pathology. Half our cohort saw an occupational therapist during the first 3 years, with 26–34% on active intervention in any year and a later peak at 3–4 years of 44% coinciding with an increase in functional skills and independence, when it is likely that families were seeking assistance for self-care skills such as toileting and dressing.

Currently, only one bone dysplasia clinic in Australia includes staffing with a dedicated physiotherapist and occupational therapist with the majority of families in Australasia sourcing services through external agencies or tertiary centre departments. However, given the frequent contact with physiotherapists, occupational therapists and speech pathologists identified in this study, increased recognition of early motor and communication delays and the restrictions for and emphasis upon careful early positioning,^{4,21,28,29} in our view, all infants with achondroplasia require assessment, parent education and management from therapists skilled in this area. Due to the low incidence of this condition, the inclusion of these professionals in multidisciplinary achondroplasia services would facilitate more reliable access for children and their families and potentially more effective services from therapists with specialised knowledge and skills in this area.

Conclusion

The Health Supervision Guidelines (1995:2005) developed for North American practices are a useful tool for guiding effective multidisciplinary management of children with achondroplasia. Our longitudinal study of Australasian children for the period 1997–2010 indicates that early access to geneticists and skilled paediatric rehabilitation physicians/paediatricians, a guideline recommendation, is high. Provision of MRI/CT and polysomnography studies appears low, reflecting a 20-year policy of conservative investigation. The numbers of children undergoing grommet insertion, tonsillectomy/adenoidectomy and CMD are

similar to results reported in previous studies. While there has been an increase in the number of children accessing speech pathology services, 40% of Australasian children are not accessing this service. Our results show a prominent role for physiotherapists and occupational therapists in the management of these children with many families accessing these professionals. With the delays in motor and postural skills and the musculoskeletal complications well documented for this population, we believe that referral for these services warrants inclusion in future guideline revisions.

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IMAGE OF THE MONTH

A 9-Year-Old Girl with a Facial Black Spot

A 9-year-old healthy girl presented in our clinic with a headache, facial erythematous rash and a black spot on her upper left eyelid. Lesions started after a day in the countryside and spread to the rest of her face. She was unable to open her left eye due to intense swelling. Two days after the appearance of the spot, she developed a non-pruritic maculopapular rash on limbs and trunk, sparing her palms and soles. Examination revealed a black crusted ulcer with intense oedema in her left eyelids, cheek and lower right eyelid (Fig. 1). The rest of the clinical exploration was normal. She had fever (39.7°C) and a blood test revealed slight thrombocytopenia ($98 \times 10^9/L$), leucocytosis with left shift (13.400/ μL ; 81% neutrophils, 12% lymphocytes, 5% monocytes, 3% eosinophils), 11.6 g/dL of haemoglobin, elevated reactive C protein (48 μ/L) and erythrocyte sedimentation rate (32 mm/h). The chest radiograph was unremarkable. What is the most likely diagnosis? (for answer, see page 455).

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Fig. 1 Black crusty spot on the left upper eyelid with local swelling.