



Dr. med. Alessandra Ferrarini

Specialist in paediatrics and specialist in medical genetics, FMH member

Consultant Medisyn Switzerland

Lecturer at the Faculty of Medicine USI Lugano

Languages

IT,FR,EN

Work Experience

- since 2024 **Paediatrician and independent clinical geneticist, at Clinica Sant'Anna, Sorengo and consultant at Medisyn, Switzerland**
- since 2022 **Accreditation at Swiss Medical Network**
- since 2021 **Lecturer at Università della Svizzera Italiana (USI)**
- 2020–2024 Paediatrician and independent clinical geneticist, at Centro Medico, Stazione, Lugano
- 2019–2022 Head Physician, Head of Medical Genetics Service at Ospedale Italiano di Lugano (IOSI-OIL)
- 2013–2018 Chief Resident Physician and then Head of Service, Paediatrics Service (IPSI) San Giovanni Hospital, Bellinzona

Education

- 2013 Resident Assistant Physician, Medical Genetics Service, Necker-Enfants Malades Hospital (Paris, France); activities at the outpatient clinics specialising in dysmorphology, genetic skeletal diseases, neurogenetic and mitochondrial diseases; head Prof Dr. med. A. Münnich
- 2011–2013 Chief physician, Integrated Service Mendrisio and Bellinzona Hospitals (Switzerland); responsible Prof Dr. med. M. G. Bianchetti and Prof. Dr. med. G.P. Ramelli Chief Consultant Physician, Integrated Service Mendrisio and Bellinzona Hospitals (Switzerland); responsible for Prof Dr. med. M. G. Bianchetti and Prof. Dr. med. G.P. Ramelli.
- 2009–2011 Assistant physician, Paediatrics Service, CHUV-Lausanne (Switzerland); responsible Prof. Dr. med. S. Fanconi
- 2007–2009 Assistant physician, Medical Genetics Service, CHUV-Lausanne (Switzerland); responsible Prof. Dr. med. J. Beckmann.
- 2004–2007 Assistant physician, Integrated Paediatrics Service, Mendrisio and Bellinzona Hospitals (Switzerland); responsible Prof. Dr. med. M.G. Bianchetti and Prof. Dr. med. G.P. Ramelli
- 2003–2004 Fellow physician, Department of Paediatrics, Medical Genetics Service, University Paediatric Clinic "G. e D. De Marchi", Milan (Italy), responsible Dr. med. A. Selicorni
- 2001–2003 Medical fellow, Department of Paediatrics, Paediatric Oncology Service, University Paediatric Clinic "G. e D. De Marchi", Milan (Italy), responsible Prof. Dr. med. V. Carnelli

1996–2001 University of Milan (Italy), Faculty of Medicine.
Diploma of medical surgeon. Title of thesis: "Interferon therapy of angiomas in paediatric age".

Memberships

since 2012 FMH Title Medical Genetics
since 2011 FMH Title in Paediatrics

Profile

SCHOLARSHIPS - SPONSORSHIPS

2003-2004: Scholarship contributed by associations of parents with children suffering from rare diseases (Cornelia de Lange National Voluntary Association; Italian Wolf-Hirschhorn Syndrome Association) for research and assistance activities.

2004: Scholarship contributed by the association AIDWEB.org-ONLUS, for collaboration in the creation of the AIDWEB website for rare diseases.

2013: Scholarship contributed by Telethon Suisse, for a clinical collaboration with the Medical Genetics Service of the Necker-Enfants Malades hospital (Paris, France).

1.11.2012: Creation of the Pediatric Rare Diseases Fund for diagnostic and research purposes in the field of rare children's diseases (subsidised by the Kiwanis Lugano Foundation for the first year of activity).

SUPERVISION OF MASTER'S THESES FOR OBTAINING A MEDICAL DEGREE

1. Ilaria Parenti (University of Milan, 2016)
2. Sabrina Mueller (University of Basel, 2017)
3. Giogia Pellanda (University of Basel, 2017)
4. Luca Jermini (University of Basel, 2018)
5. Gabriel Bronz (University of Bern, 2018)

REVIEWER ACTIVITIES FOR SCIENTIFIC JOURNALS

European Journal of Pediatrics - Human Genetics - The Journal of Pediatrics

PARTICIPATION IN INTERNATIONAL RESEARCH CONSORTIA

1. Consortium 16p11.2:

- 16p11.2 Locus modulates response to satiety before the onset of obesity. Int J Obes 2016 40:870-876.

- Chromosomal contacts connect loci associated with autism, BMI and head circumference phenotypes. Mol Psychiatry 2016; 22:836-849.

- Defining the effect of the 16p11.2 duplication on cognition, behaviour, and medical comorbidities. JAMA Psychiatry 2016; 73:20-30.

2. Consortium 15q11.2:

- Estimating the effect size of the 15q11.2 BP1-BP3 deletion and its contribution to neurodevelopmental symptoms: recommendation for practise. J Med Genet 2019;56:701-710.

Publications

BIBLIOGRAPHY (peer-reviewed scientific journals)

Hirsch factor: 10 (ISI; 8 June 2020)

- ORIGINAL ARTICLES (AND SYSTEMATIC REVIEWS)

1. Zollino M, Lecce R, Selicorni A, Murdolo M, Mancuso I, Marangi G, Zampino G, Garavelli L, **Ferrarini A**, Rocchi M, Opitz JM, Neri G. A double cryptic chromosome imbalance is an important factor to explain phenotypic variability in Wolf-Hirschhorn syndrome. *Eur J Hum Genet* 2004;12:797-804.
2. Bartoli F, Martinez JM, **Ferrarini A**, Recaldini E, Bianchetti MG. Poor adherence to the prophylactic use of vitamin D3 in Switzerland. *J Pediatr Endocrinol Metab* 2006;19:281-282.
3. Robbiani JH, Simonetti G, Crosazzo L, **Ferrarini A**, Pronzini F, Bianchetti MG. False positive dipstick for urinary blood in childhood. *J Nephrol* 2006;19:605-606.
4. Van der Aa N, Rooms L, Vandeweyer G, van den Ende J, Reyniers E, Fichera M, Romano C, Delle Chiaie B, Mortier G, Menten B, Destrée A, Maystadt I, Männik K, Kurg A, Reimand T, McMullan D, Oley C, Brueton L, Bongers EM, van Bon BW, Pfund R, Jacquemont S, **Ferrarini A**, Martinet D, Schrandt-Stumpel C, Stegmann AP, Frints SG, de Vries BB, Ceulemans B, Kooy RF. Fourteen new cases contribute to the characterization of the 7q11.23 microduplication syndrome. *Eur J Med Genet* 2009;52:94-100.
5. **Ferrarini A**, Osterheld MC, Vial Y, de Viragh PA, Cotting J, Martinet D, Beckmann JS, Fellmann F. Familial occurrence of an association of multiple intestinal atresia and choanal atresia: a new syndrome? *Am J Med Genet A* 2009;149A:2661-2665.
6. Walters RG, Jacquemont S, Valsesia A, de Smith AJ, Martinet D, Andersson J, Falchi M, Chen F, Andrieux J, Lobbens S, Delobel B, Stutzmann F, El-Sayed Moustafa JS, Chèvre JC, Lecoeur C, Vatin V, Bouquillon S, Buxton JL, Boute O, Holder-Espinasse M, Cuisset JM, Lemaitre MP, Ambresin AE, Brioschi A, Gaillard M, Giusti V, Fellmann F, **Ferrarini A**, Hadjikhani N, Champion D, Guilmatre A, Goldenberg A, Calmels N, Mandel JL, Le Caignec C, David A, Isidor B, Cordier MP, Dupuis-Girod S, Labalme A, Sanlaville D, Béri-Dexheimer M, Jonveaux P, Leheup B, Ounap K, Bochukova EG, Henning E, Keogh J, Ellis RJ, Macdermot KD, van Haelst MM, Vincent-Delorme C, Plessis G, Touraine R, Philippe A, Malan V, Mathieu-Dramard M, Chiesa J, Blaumeiser B, Kooy RF, Caiazzo R, Pigeyre M, Balkau B, Sladek R, Bergmann S, Mooser V, Waterworth D, Reymond A, Vollenweider P, Waeber G, Kurg A, Palta P, Esko T, Metspalu A, Nelis M, Elliott P, Hartikainen AL, McCarthy MI, Peltonen L, Carlsson L, Jacobson P, Sjöström L, Huang N, Hurles ME, O'Rahilly S, Farooqi IS, Männik K, Jarvelin MR, Pattou F, Meyre D, Walley AJ, Coin LJ, Blakemore AI, Froguel P, Beckmann JS. A new highly penetrant form of obesity due to deletions on chromosome 16p11.2. *Nature* 2010;463:671-675.

7. Jacquemont S, Reymond A, Zufferey F, Harewood L, Walters RG, Kutalik Z, Martinet D, Shen Y, Valsesia A, Beckmann ND, Thorleifsson G, Belfiore M, Bouquillon S, Campion D, de Leeuw N, de Vries BB, Esko T, Fernandez BA, Fernández-Aranda F, Fernández-Real JM, Gratacòs M, Guilmatre A, Hoyer J, Jarvelin MR, Kooy RF, Kurg A, Le Caignec C, Männik K, Platt OS, Sanlaville D, Van Haelst MM, Villatoro Gomez S, Walha F, Wu BL, Yu Y, Aboura A, Addor MC, Alembik Y, Antonarakis SE, Arveiler B, Barth M, Bednarek N, Béna F, Bergmann S, Beri M, Bernardini L, Blaumeiser B, Bonneau D, Bottani A, Boute O, Brunner HG, Cailley D, Callier P, Chiesa J, Chrast J, Coin L, Coutton C, Cuisset JM, Cuvellier JC, David A, de Freminville B, Delobel B, Delrue MA, Demeer B, Descamps D, Didelot G, Dieterich K, Disciglio V, Doco-Fenzy M, Drunat S, Duban-Bedu B, Dubourg C, El-Sayed Moustafa JS, Elliott P, Faas BH, Faivre L, Faudet A, Fellmann F, **Ferrarini A**, Fisher R, Flori E, Forer L, Gaillard D, Gerard M, Gieger C, Gimelli S, Gimelli G, Grabe HJ, Guichet A, Guillin O, Hartikainen AL, Heron D, Hippolyte L, Holder M, Homuth G, Isidor B, Jaillard S, Jaros Z, Jiménez-Murcia S, Helas GJ, Jonveaux P, Kaksonen S, Keren B, Kloss-Brandstätter A, Knoers NV, Koolen DA, Kroisel PM, Kronenberg F, Labalme A, Landais E, Lapi E, Layet V, Legallic S, Leheup B, Leube B, Lewis S, Lucas J, MacDermot KD, Magnusson P, Marshall C, Mathieu-Dramard M, McCarthy MI, Meitinger T, Mencarelli MA, Merla G, Moerman A, Mooser V, Morice-Picard F, Mucciolo M, Nauck M, Ndiaye NC, Nordgren A, Pasquier L, Petit F, Pfundt R, Plessis G, Rajcan-Separovic E, Ramelli GP, Rauch A, Ravazzolo R, Reis A, Renieri A, Richart C, Ried JS, Rieubland C, Roberts W, Roetzer KM, Rooryck C, Rossi M, Saemundsen E, Satre V, Schurmann C, Sigurdsson E, Stavropoulos DJ, Stefansson H, Tengström C, Thorsteinsdóttir U, Tinahones FJ, Touraine R, Vallée L, van Binsbergen E, Van der Aa N, Vincent-Delorme C, Visvikis-Siest S, Vollenweider P, Völzke H, Vulto-van Silfhout AT, Waeber G, Wallgren-Pettersson C, Witwicki RM, Zvolinski S, Andrieux J, Estivill X, Gusella JF, Gustafsson O, Metspalu A, Scherer SW, Stefansson K, Blakemore AI, Beckmann JS, Froguel P. Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. *Nature* 2011; 478: 97-102.
8. **Ferrarini A**, Auteri-Kaczmarek A, Pica A, Boesch N, Heinimann K, Schafer SC, Vesnaver-Megalo S, Cina V, Beckmann JS, Monnerat C. Early occurrence of lung adenocarcinoma and breast cancer after radiotherapy of a chest wall sarcoma in a patient with a de novo germline mutation in TP53. *Fam Cancer* 2011; 10: 187-192.
9. Gervasini C, Russo S, Cereda A, Parenti I, Masciadri M, Azzollini J, Meli D, Aravena T, Doray B, **Ferrarini A**, Garavelli L, Selicorni A, Larizza L. Cornelia de Lange individuals with new and recurrent *SMC1A* mutations enhance delineation of mutation repertoire and phenotypic spectrum. *Am J Med Genet A* 2013; 161A: 2909-2919.
10. Lava SAG, Simonetti GD, **Ferrarini A**, Ramelli GP, Bianchetti MG. Regional differences in symptomatic fever management among pediatricians in Switzerland: The results of a cross-sectional Web-based survey. *Br J Clin Pharmacol* 2013; 75: 236-243.
11. **Ferrarini A**, Lava SAG, Simonetti GD, Ramelli GP, Bianchetti MG; on behalf of the Swiss Italian Society of Pediatrics. Influenzavirus B-associated acute benign myalgia cruris: An outbreak report and review of the literature. *Neuromuscul Disord* 2014;24:342-346.
12. Uestuener P, **Ferrarini A**, Santi M, Mardegan C, Bianchetti MG, Simonetti GD, Milani GP, Lava SAG. Taste acceptability of pulverized brand-name and generic drugs containing amlodipine or candesartan. *Int J Pharm* 2014;468:196-198.
13. Siegenthaler GM, Rizzi M, Bettinelli A, Simonetti GD, **Ferrarini A**, Bianchetti MG. Ureteral or vesical involvement in Henoch-Schönlein syndrome: a systematic review of the literature. *Pediatr Nephrol* 2014; 29: 235-239.

14. Ostini A, Ramelli GP, Mainetti C, Bianchetti MG, **Ferrarini A**. Recurrent Finkelstein-Seidlmayer disease in four first-degree relatives. *Acta Derm Venereol* 2015;28:622-623.
15. Jackson CB, Bauer MF, Schaller A, Kotzaeridou U, Ferrarini A, Hahn D, Chehade H, Barbey F, Tran C, Gallati S, Haeberli A, Eggimann S, Bonafé L, Nuoffer JM. A novel mutation in BCS1L associated with deafness, tubulopathy, growth retardation and microcephaly. *Eur J Pediatr* 2016;175:517-525.
16. Capoferri G, Milani GP, Ramelli GP, **Ferrarini A**, Bianchetti MG. Sporadic acute benign calf myositis: Systematic literature review. *Neuromuscul Disord* 2018;28:443-449.
17. O'Donnell-Luria AH, Pais LS, Faundes V, Wood JC, Sveden A, Luria V, Abou Jamra R, Accogli A, Amburgey K, Anderlid BM, Azzarello-Burri S, Basinger AA, Bianchini C, Bird LM, Buchert R, Carre W, Ceulemans S, Charles P, Cox H, Culliton L, Currò A; Deciphering Developmental Disorders (DDD) Study, Demurger F, Dowling JJ, Duban-Bedu B, Dubourg C, Eiset SE, Escobar LF, **Ferrarini A**, Haack TB, Hashim M, Heide S, Helbig KL, Helbig I, Heredia R, Héron D, Isidor B, Jonasson AR, Joset P, Keren B, Kok F, Kroes HY, Lavillaureix A, Lu X, Maas SM, Maegawa GHB, Marcelis CLM, Mark PR, Masruha MR, McLaughlin HM, McWalter K, Melchinger EU, Mercimek-Andrews S, Nava C, Pendziwiat M, Person R, Ramelli GP, Ramos LLP, Rauch A, Reavey C, Renieri A, Rieß A, Sanchez-Valle A, Sattar S, Saunders C, Schwarz N, Smol T, Srour M, Steindl K, Syrbe S, Taylor JC, Telegrafi A, Thiffault I, Trauner DA, van der Linden H Jr, van Koningsbruggen S, Villard L, Vogel I, Vogt J, Weber YG, Wentzensen IM, Widjaja E, Zak J, Baxter S, Banka S, Rodan LH. Heterozygous variants in KMT2E cause a spectrum of neurodevelopmental Disorders and epilepsy. *Am J Hum Genet* 2019;104:1210-1222.
18. [Ferrarini A](#), [Benetti C](#), [Camozzi P](#), [Ostini A](#), [Simonetti GD](#), [Milani GP](#), [Bianchetti MG](#), [Lava SA](#). Acute hemorrhagic edema of young children: a prospective case series. *Eur J Pediatr* 2016;175:557-561.
19. Ostini A, Simonetti GD, Pellanda G, Bianchetti MG, **Ferrarini A**, Milani GP. Familial Henoch-Schönlein syndrome. *J Clin Rheumatol* 2016;22:80-81.
20. Bronz G, Gabriel H, Lava SAG, Ramelli GP, Luedeke M, Biskup S, Mainetti C, **Ferrarini A**. Whole exome sequencing revealed a candidate gene for Finkelstein-Seidlmayer disease. *Am J Pediatr* 2019;5:196-199.
21. Rinoldi PO, Milani GP, Bianchetti MG, **Ferrarini A**, Ramelli GP, Lava SAG. Acute hemorrhagic edema of young children: open questions and perspectives. *Int J Dermatol Skin Care* 2019;1:63-67.
22. O'Donnell-Luria AH, Pais LS, Faundes V, Wood JC, Sveden A, Luria V, Abou Jamra R, Accogli A, Amburgey K, Anderlid BM, Azzarello-Burri S, Basinger AA, Bianchini C, Bird LM, Buchert R, Carre W, Ceulemans S, Charles P, Cox H, Culliton L, Currò A; Deciphering Developmental Disorders (DDD) Study, Demurger F, Dowling JJ, Duban-Bedu B, Dubourg C, Eiset SE, Escobar LF, **Ferrarini A**, Haack TB, Hashim M, Heide S, Helbig KL, Helbig I, Heredia R, Héron D, Isidor B, Jonasson AR, Joset P, Keren B, Kok F, Kroes HY, Lavillaureix A, Lu X, Maas SM, Maegawa GHB, Marcelis CLM, Mark PR, Masruha MR, McLaughlin HM, McWalter K, Melchinger EU, Mercimek-Andrews S, Nava C, Pendziwiat M, Person R, Ramelli GP, Ramos LLP, Rauch A, Reavey C, Renieri A, Rieß A, Sanchez-Valle A, Sattar S, Saunders C, Schwarz N, Smol T, Srour M, Steindl K, Syrbe S, Taylor JC, Telegrafi A, Thiffault I, Trauner DA, van der Linden H Jr, van Koningsbruggen S, Villard L, Vogel I, Vogt J, Weber YG, Wentzensen IM, Widjaja E, Zak J, Baxter S, Banka S, Rodan LH. Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. *Am J Hum Genet*. 2019;104(6):1210-1222.

23. Pellanda F, Lava SAG, Milani GP, Bianchetti MG, **Ferrarini A**, Vanoni F. Immune deposits in skin vessels of patients with acute hemorrhagic edema of young children: a systematic literature review. *Pediatr Dermatol* 2020;37:120-123.
24. Malhotra A, **Ferrarini A** et al. De novo missense variants in LMBRD2 are associated with developmental and motor delays, brain structure abnormalities and dysmorphic features. *J Med Genet* 2020.
25. Lopergolo D, Privitera F, Castello G, Lo Rizzo C, Mencarelli MA, Pinto AM, Ariani F, Curro' A, Lamacchia V, Canitano R, Vaghi E, **Ferrarini A**, Baltodano GM, Lederer D, Van Maldergem L, Serrano M, Pineda M, Fons-Estupina MDC, Van Esch H, Breckpot J, Kumps C, Callewaer B, Mueller S, Rameli GP, Armstrong J, Renieri A, Mari F. IQSEC2 disorder: a new entity or a Rett spectrum continuum? *Clin Genet* 2021;99(3):462-474.
26. Betti C, Camozzi P, Gennaro V, Bianchetti MG, Scoglio M, Simonetti GD, Milani GP, Lava SAG, **Ferrarini A**. Atypical bacterial pathogens and small-vessel leukocytoclastic vasculitis of the skin in children: systematic literature review. *Pathogens* 2021;10(1):31-.

- NARRATIVE REVIEWS

27. Santoro L, **Ferrarini A**, Crosazzo L, Bianchetti MG. Primary vesicoureteric reflux and reflux nephropathy - new insights. *Curr Pediatr Rev* 2006;2:173-176.
28. Simonetti GD, Santoro L, **Ferrarini A**, Crosazzo-Frascini L, Fossali E, Bianchetti MG. Systemic hypertension and proteinuria in childhood chronic renal parenchymal disease. Role of antihypertensive drug management. *Paediatr Drugs* 2007;9:413-418.
29. Bucher BS, **Ferrarini A**, Weber N, Bullo M, Bianchetti MG, Simonetti GD. Primary hypertension in childhood. *Curr Hypertens Report* 2013;15:444-452.
30. **Ferrarini A**, Jacquemont S, Beck Popovic M, Bonafé L, Martinet D. Puce à ADN: pourquoi et pour qui. *Rev Med Suisse* 2010;237:390-396.
31. Lava SAG, Simonetti GD, Bianchetti AA, **Ferrarini A**, Bianchetti MG. Prevention of vitamin D insufficiency in Switzerland: A never-ending story. *Int J Pharm* 2013;457:353-356.
32. **Ferrarini A**, Bianchetti AA, Fossali EF, Faré PB, Simonetti GD, Lava SAG, Bianchetti MG. What can we do to make antihypertensive medications taste better for children? *Int J Pharm* 2013;457:333-336.

- LETTERS - CASE REPORT

33. **Ferrarini A**, Selicorni A, Cagnoli G, Zollino M, Lecce R, Chines C, Battaglia A. Distinct facial dysmorphism, pre and postnatal growth retardation, microcephaly, seizures, mental retardation and hypotonia. *Ital J Pediatr* 2003;29:393-397.
34. Selicorni A, **Ferrarini A**, Cagnoli G, Fratoni A, Bottigelli M, Milani D. Additional case of Tsukahara's syndrome or new syndrome: further delineation of the association of microcephaly and radio-ulnar synostosis *Am J Med Genet* 2005;132A:189-190.
35. **Ferrarini A**, Bottigelli M, Milani D, Cagnoli G, Selicorni A. Two new cases of Barraquer-Simons syndrome. *Am J Med Gen* 2004;126A:427-429.
36. **Ferrarini A**, Baggi M, Flückiger R, Bianchetti MG. Intraoperative anaphylaxis to a chlorhexidine polymer in childhood. *Paediatr Anaesth* 2006;16:705.

37. Baldassarre E, Mendoza Sagaon M, **Ferrarini A**, Bianchetti MG. Severe systemic adverse reaction to proton pump inhibitors in an infant. *Pediatr Pulmonol* 2007;42:563-564.
38. **Ferrarini A**, Blumberg D, Cortesi C, Bianchetti MG. Henoch-Schönlein syndrome and pertussis. *Pediatr Infect Dis J* 2007;26:661.
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40. **Ferrarini A**, Ramelli GP, Bianchetti MG, Hedman J, Sharathkumar A, Shapiro A, Bourland C. Index of suspicion. *Pediatr Rev* 2009;30:479-485.
41. Capobianco S, Lava SA, Bianchetti MG, Martinet D, Belfiore M, Ramelli GP, **Ferrarini A**. Chromosomal microarray among children with intellectual disability: a useful diagnostic tool for the clinical geneticist. *Dev Med Child Neurol* 2014;56:290.
42. **Ferrarini A**, Gaillard M, Guerry F, Ramelli GP, Heidi F, Keddache CV, Wieland I, Beckmann JS, Jaquemont S, Martinet D. Potocki-Shaffer deletion encompassing ALX4 in a patient with frontonasal dysplasia phenotype. *Am J Med Genet A*. 2014;164:346-352.
43. Pellanda G, Lava SA, **Ferrarini A**, Ramelli GP. High prevalence of pathologic copy number variants detected by chromosomal microarray in Swiss-Italian children with autism spectrum disorders. *Eur J Paediatr Neurol* 2015;19:387-387.
44. **Ferrarini A**, Milani GP, Bianchetti MG, Lava SAG. Acute hemorrhagic edema of infancy associated with Cocksackie virus infection. *Arch Pédiatr* 2018;25:244.

Bronz G, Leoni-Foglia C, Lava SG, Simonetti GD, **Ferrarini A**. Pathologic fracture revealed a rare syndromic form of genetic lipodystrophy. *Clin Dymorphol* 2020;29:53-60.

Accreditation

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Specialties

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