|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Table 3.** The reported cases and *KITLG* mutations of familial progressive hyper- and hypopigmentation to date | Reference | [1] | [2,3] | [2,3] | [2,4] | [2] | [5] | [5] | [6] | [7] | [8] | This study | This study |
| Amino acid changes | p.Asn36Ser | p.Asn36Ser | p.Asn36Ser | p.Val33Ala | p.Thr34Pro | p.Thr34Asn | p.Val37Gly | p.Thr34Ile | p.Thr34Ile | p.Glu113Lys | p.Thr34Ile | p.Asn35Ile |
| Variant type | Missense | Missense | Missense | Missense | Missense | Missense | Missense | Missense | Missense | Missense | Missense | Missense |
| Mutation | c.107A>G | c.107A>G | c.107A>G | c.98T>C | c.100A>C | c.101C>A | c.110T>G | c.101C>T | c.101C>T | c.337G＞A | c.101C>T | c.104A>T |
| Exon | 2 | 2 | 2 | 2 | 2 | 2 | 2 | 2 | 2 | 4 | 2 | 2 |
| Cancer |  |  |  |  |  | pharyngeal cancer; melanoma | papillary thyroid cancer |  |  |  |  |  |
| Height |  |  |  |  |  | below average |  |  |  |  |  |  |
| Clinical manifestation | diffuse hyperpigmentation | diffuse hyperpigmentation, hypopigmented ash-leaf-like lesions, cafe´-au-lait macules, lentigines | diffuse hyperpigmentation, confetti-like hypopigmented macules, cafe´-au-lait macules, lentigines | hyper- and hypopigmented macules/patches, cafe´-au-lait macules | diffuse hyperpigmentation, with scattered cafe´-au-lait macules, lentigines, small hypopigmented spots and vitiligo | hyper- and hypopigmented macules, cafe´-au-lait-like macules and large irregular hypopigmented macules, unusually sparse lateral eyebrows | generalized freckling and hyperpigmentation, lipo-oedema of the legs, unusually sparse lateral eyebrows | diffuse hyperpigmentation intermixed with CALS, lentigines, hypopigmented macules and spots | diffuse hyperpigmentation with irregular café-au-lait and café noir spots, ash leaf-like macules, hypopigmented patches and intense hyperpigmentation | irregularly shaped asymptomatic hyper- and hypopigmented macules | diffuse hyper- hypopigmentation and vast café-au-lait like lesions | generalized hyper- and hypopigmentation |
| Nationality | Chinese | Germany | Germany | French Canadian origin | Danish | British | British | Chinese | Turkey | Japanese | Chinese | Chinese |
| Patients | Family 1 | Family 2 | Family 3 | Family 4 | Family 5 | Family 6 | Family 7 | Sporadic case 1 | Sporadic case 2 | Family 8 | Sporadic case 3 | Family 9 |

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