**Table SII. Clinical features in 4 conditions caused by mutations of *PDGFRB* (adapted from (8))**

| Clinical manifestations | Familial infantile myofibromatosis | Penttinen syndrome | Syndrome described by Takenouchi et al. (9) | Idiopathic basal ganglia calcification |
|---|---|---|---|---|
| Skin | Solitary or multicentric myofibromas | Lipoatrophy, hyperkeratotic lesions, thin skin | Thin skin, 1 individual with solitary myofibroma | – |
| Brain | – | – | Neuro-behavioural manifestations<br>Periventricular white matter lesions | Dystonia, ataxia, neuro-behavioural manifestations, basal ganglia calcifications |
| Skeletal | – | Scoliosis | Acro-osteolysis, osteopaenia | – |
| Dysmorphy | – | + | – | – |
| Overgrowth | – | – | + | – |
| Ocular | – | NS | NS | – |

Major clinical findings reported in familial IM, Penttinen syndrome, overgrowth syndrome (9) and idiopathic basal ganglia calcification.
+: positive; –: negative; NS: not assessed.